

Klippel–Trenaunay Syndrome: A Rare Case Presenting in a 5-Year-Old Girl

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ABSTRACT

A 5-year-old child presented with progressive right calf swelling for 4 years and a year history of pain at the same site. There was no skin discoloration. Doppler ultrasound scan and venography of the affected leg showed soft-tissue gigantism and anomalous venous drainage of the right leg with multiple tortuous varicose veins and venous aneurysm. The deep right calf veins were anomalous but ipsilateral popliteal and femoral veins were preserved. The arterial system of the right lower limb was preserved; no arteriovenous malformation was seen, and no limb length discrepancy or evidence of bone involvement was observed. The diagnosis of Klippel–Trenaunay syndrome was made based on the above clinical and radiological features. After due consultations with the pediatrician and orthopedic surgeon, she was commenced on analgesic and application of graded compression stockings, and she's doing well.

Key words: Doppler ultrasound; gigantism; Klippel–Trenaunay syndrome; port-wine stain; venography

Introduction

Klippel–Trenaunay Syndrome (KTS) is a congenital vascular disorder of unknown etiology. It is a variant of Von Hippel–Lindau disease, a rare myelocutaneous disorder in which a vascular malformation of the spinal cord and meninges is associated with vascular nevus within the area of skin innervated by the involved spinal segment and enlargement of the affected extremity.^[1]

KTS is characterized by a triad of port-wine birthmark due to capillary malformation, soft-tissue or/and bone hypertrophy (localized gigantism), and varicose veins or venous malformation. It is considered an angio-osteo-hypertrophic syndrome.^[2] The diagnosis of KTS is usually made when any two of the three features are present. Port-wine nevus (capillary malformation) may be absent in the atypical form.^[3] The rarity of the case

particularly in Nigeria, where no case of such has been reported, prompted this report.

Case Report

E G is a 5-year-old girl who presented to our health facility (Federal Staff Medical Centre, Abuja) with 4 years history of swelling of the right calf and a year history of pain. The abnormal swelling was first noticed at 1-year of age when the mother observed that the right calf was larger than that of the left. There was no preceding trauma to the site. There was also no skin discoloration or gait disturbance.

The swelling was said to have progressively increased in size with no untoward effect. Patient, however, developed pain at the same site about a year ago. The character of the pain

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could not be elicited but was said to be aggravated by walking, and it progressively worsened about 3 months prior to the presentation, making her to limp. No medical or unorthodox intervention had been sought.

There was no history of abnormal bleeding from any orifice, and no headache or a history of convulsion.

No similar presentation in any of the siblings. She is the 3rd of her parents' three children in a monogamous family.

On examination, she was a preschool age child not in any painful distress, afebrile, acynosed, not pale, anicteric, but had vernal conjunctivitis. No significant peripheral lymph node enlargement was noted.

Musculoskeletal system examination showed an obvious hypertrophy of the right calf with a few varicose veins draining upward. It extended from 1cm below the popliteal fossa to 2 cm above the ankle joint measuring 28.5 cm at the widest circumference, 12 cm from the patella. The left measured 25.5 cm from the same landmark [Figure 1]. There was associated moderate tenderness, but the patient had full range of movement across the right knee and ankle joints.

There was no skin discoloration, apparent or real shortening of the limbs and no bruit over the swelling.

All peripheral pulses were palpable with the radial pulse being 80 beats/min, regular and full volume. The blood pressure was 90/65 mmHg, and the apex beat was localized at 5th left intercostals space mid clavicular line. The remaining systems were essentially preserved.

The full blood count done showed normal findings and the child genotype was HbAA.



Figure 1: A pictorial image showing hypertrophied soft-tissue of the right calf (soft-tissue gigantism)

Ultrasounds scan Figure 2 shows hypertrophy of the soft-tissue of the right calf (soft-tissue gigantism) with dilatation and abnormal branching of the superficial veins of the affected calf. The dilated superficial veins show normal spectral waveform with no evidence of arterIALIZATIONS seen. The abdominal viscera were essentially normal on ultrasound scan.

The right leg venogram [Figures 3 and 4] shows patternless, tortuous, and dilated superficial veins of the calf communicating with the small saphenous vein and anomalous deep venous system of the same leg at variable levels. Multiple tortuous varicose veins were demonstrated presumably due to incompetent valveless collateral venous channels. The deep venous system of the right leg appears anomalous with multiple focal dilatations seen along its course (venous aneurysm), before draining into the popliteal vein. The popliteal and femoral veins are unremarkable.

In view of the enumerated clinical features coupled with radiological findings, a diagnosis of KTS was made.

The patient's mother was counseled on the nature of the disease. Orthopedic surgeon reviewed the patient and she's being managed with graded compressive stockings. She is currently on regular follow-up at the pediatric outpatient clinic of the hospital.

Discussion

KTS also known as angio-osteo-hypertrophic syndrome was first described by two French doctors, Klippel Marcus and Trenaunay Philip in 1900.^[4] It is a rare disorder with an incidence of 3–5/1000,000.^[5] The syndrome is characterized by the triad of vascular malformation (port-wine stain), venous varicosities/malformation, and soft-tissue and/or bony hypertrophy (focal gigantism).^[4] In some cases, port-wine stains may be absent. Such cases are very rare and may be classified as atypical KTS.^[6] This is the case scenario in this index case.

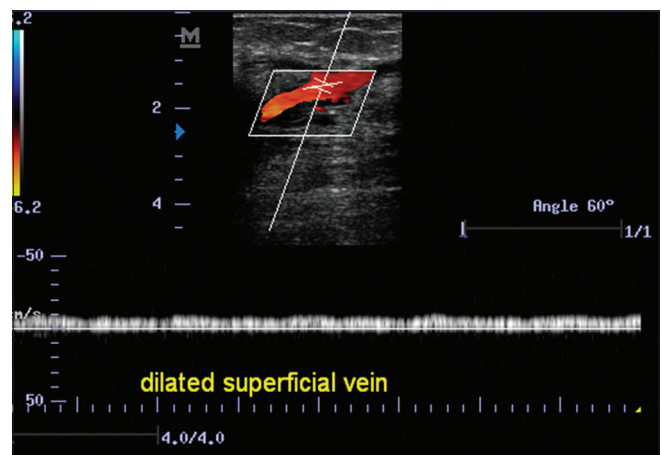


Figure 2: Duplex Doppler ultrasound image of the right leg showing dilated superficial vein with normal venous spectral waveform

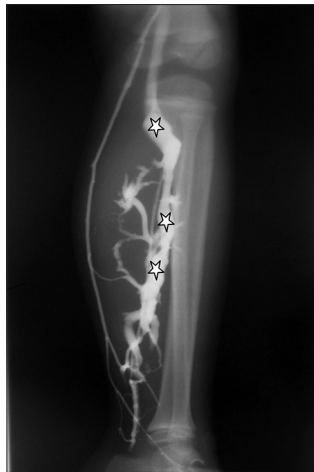


Figure 3: Right leg venogram showing patternless, tortuous and dilated superficial veins of the calf communicating with the small saphenous vein and deep venous system of the same leg at variable levels. The deep venous system appears anomalous with multiple focal dilatations seen along its course (venous aneurysm-point stars), before draining into the popliteal vein

KTS occurs sporadically and shows no particular racial, gender or geographic predilection.^[7] It affects the skin, veins, lymphatic system, bone and soft-tissue of an extremity. If the cutaneous hemangiomas are large enough, it may cause sequestration of platelets, leading to Kasabach-Merritt syndrome (consumptive coagulopathy).^[8,9] The hemangiomas in the index case is small which explains why the patient had no disseminated intravascular coagulopathy at presentation and her platelet count was within normal limits. Furthermore, the varicosities associated with this syndrome can produce venous stasis consequently leading to pain, bleeding, thrombophlebitis, and pulmonary embolism. The majority of these features, other than pain, were not found in the patient at presentation. This may be explained by the age of the patient, and it's, therefore, imperative to serially assess her for the possibility of these complications at subsequent follow-up, and all efforts shall be geared toward averting them.

The limb length discrepancy observed in many patients with KTS was not observed in the index case, but the girth of the calf of the affected limb is larger than the contralateral side. Atrophy has been reported in few cases. The possibilities of these features occurring at a later date cannot be ruled-out absolutely.

KTS is usually unilateral with lower limb involvement in about 95% of cases, and upper limb in 5%.^[10] This is in conformity with the index case where the right lower limb is affected, sparing other limbs. Macrodactyly, syndactyly, polydactyly, and oligodactyly are some of the limb abnormalities seen in some cases of KTS but are not the case scenario in the index case.

Some patients with KTS have also been found to present with gastrointestinal (GI) bleeding, esophageal varices,



Figure 4: Right leg venogram showing multiple tortuous varicose veins (point star) presumably due to incompetent valveless collateral venous channels

genitourinary bleeding as a result of urinary bladder varices, and splenic hemangiomas.^[6,11] These features were ruled out in the index case both clinically and radiologically.

Although the cause of KTS is still unknown, it is hypothesized, that it is caused by mesodermal abnormality during fetal development.^[12] Furthermore, Berry *et al.*^[13] reviewed 49 cases of KTS, and all were found to be sporadic. They speculated that the disorder may be due to a somatic mutation for a factor critical to vasculogenesis and angiogenesis in embryonic development.

Differentiating KTS from a closely related differential is imperative. Parkes Weber syndrome is similar to KTS except that an arteriovenous malformation (AVM) occurs in association with a cutaneous capillary malformation and skeletal or soft-tissue hypertrophy.^[14] AVM was ruled out in the index case on radiological grounds (venography and venous Doppler of the affected limb), which further confirms the diagnosis.

No single treatment protocol is applicable to all cases of KTS. Treatment is decided on a case-by-case basis, which is usually conservative and only rarely is surgical or orthopedic intervention warranted.^[15] Available treatment options include: Analgesia for thrombophlebitis and cellulitis, antibiotics for similar reasons as above, application of graded compression stockings or pneumatic compression devices to the enlarged extremity, sclerotherapy with alcohol or foam, surgical stripping, phlebectomy, endovascular thermal ablation or rarely deep venous reconstruction, selective arterial embolization for GI bleeding, surgical resection of the bowel may be required in some cases of severe GI bleeding, splenectomy for splenic hemangioma, correction of limb length discrepancy by orthoses, and amputation in some cases.^[16,17] The index case is being managed with analgesic and application of graded compression stockings, and she's doing well.

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Conflicts of interest

There are no conflicts of interest.

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