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A case report

Klippel-Trenaunay Syndrome – An interesting syndrome in a Nigerian child

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Abstract

Klippel-Trenaunay Syndrome (KTS) is a complex and unusual congenital angio-osteo-hypertrophic disorder where affected individuals have cutaneous hemangioma, venous and lymphatic malformations, and hemihypertrophy of the soft tissue and bones of the limb, causing limb asymmetry. We report a 4-year-old child with ipsilateral hemihypertrophy and cutaneous hemangioma, where KTS diagnosis was made despite being challenging.

Keywords: Capillary malformations, Congenital disorder, Klippel-Trenaunay syndrome, Limb hypertrophy, Varicosities.

Introduction

Klippel-Trenaunay syndrome (KTS) was first described by French physicians Klippel and Trenaunay (1). It is an uncommon congenital syndrome that classically consists of a vascular malformation syndrome involving the cutaneous capillaries, veins, and lymphatics, with soft tissue hyperplasia and bone hypertrophy (1,2). These lesions first appear at birth and infancy and progress into adolescence (3). Two or three features are needed to diagnose KTS (4). It usually affects one limb, more frequently in the lower limb, and the right lower limb is more frequently affected than the left (4). Rarely does it affect the upper extremity and both lower limbs, and a case of crossed-bilateral KTS has been reported (5).

The incidence is unknown, estimated at 2– 5 in 100,000 individuals (6). It has no ethnic or sex prevalence or sex preference (7). It is a benign disease, with its severity depending on the magnitude of vascular dysplasia, visceral involvement, and complications. We report a case of KTS in a Nigerian child, which will increase awareness among physicians as its diagnosis may be challenging.

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Case presentation

A 4-year-old boy presented in our facility on account of non-painful right lower limb swelling since birth with gait instability. The right limb progressively increased, bigger than the left, and extended from the right thigh to the foot, with gait affectation. Ipsilateral buttocks, thigh, and leg hyperpigmentation were noted from birth, with a few patches at the trunk. He had no bone pain, headache, seizure, or developmental delay. He is the 3rd of three children in a polygamous family with no similar presentation in his siblings.

On examination, he had a limp right leg with minimally loose over-hanging skin folds on the plantar surface of the right foot and no differential warmth in either limb. More posteriorly, hyperpigmented patches were seen in the right buttocks to the foot [Figure 1]. There were discrepancies in the limbs' apparent and real lengths (Right– 61cm; 59cm, and left – 56cm; 51cm). Girth differences were noted in both thighs and calves (Right – 27cm; 20cm, and left – 25cm; 16cm), taken at 15cm and 10cm from the anterior superior iliac spines, respectively. The ankle circumferences were 18cm and 14cm on the right and left, respectively; however, there was a full bilateral range of movement across the joints. [Figures 2 &3]

Figure 1: Photo showing the distribution of hyperpigmentation on the right lower limb and torso and the asymmetrical thickening and length of the right lower limb.

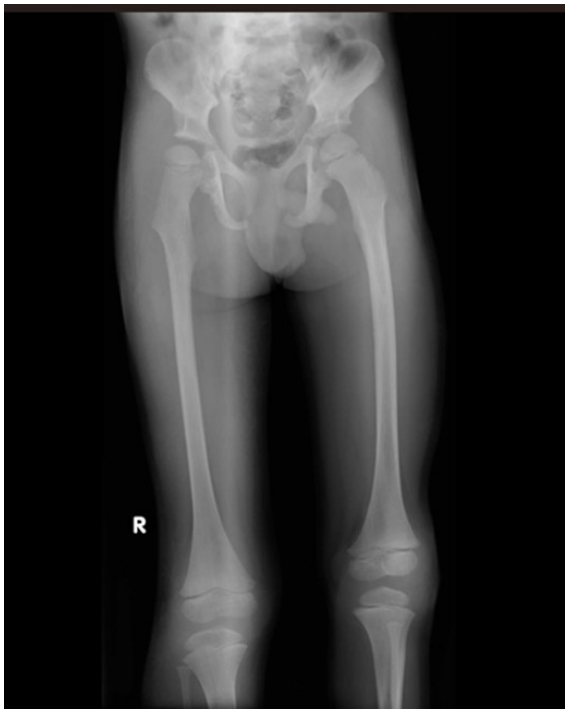

Figure 2: Anteroposterior radiographs of the femurs showing a longer right femur.

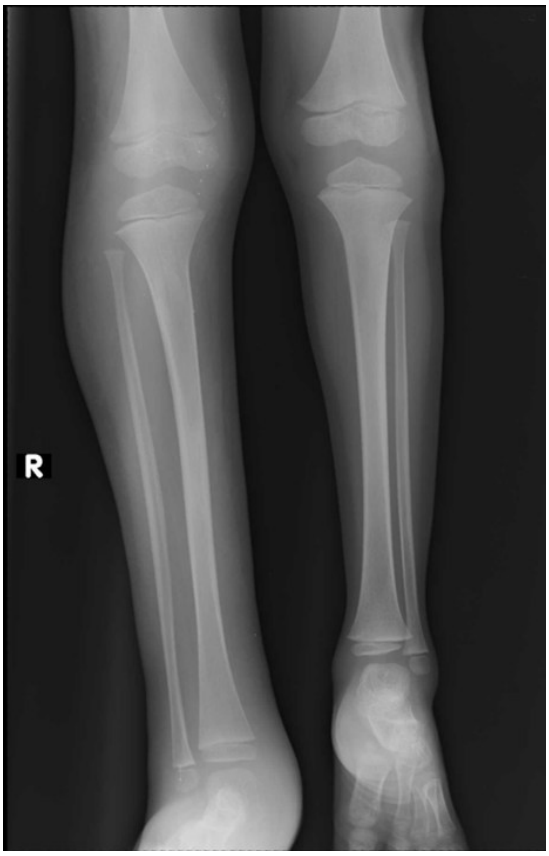

Figure 3: Anteroposterior radiograph of both tibia and fibula shows longer right tibia and fibula bones with fullness of soft tissue in the right leg.

Laboratory studies revealed iron deficiency anemia with a hematocrit of 33.7% (38.6–49.2%) and MCH of 22.9 (26–34). The white blood cells and platelets were normal. A color Doppler ultrasonography of the lower limbs showed small-sized ectatic veins at the lateral aspect of the right leg with no unusual connection to the venous systems. No deep vein thrombosis or subcutaneous edema was seen. The nonavailability of a magnetic resonance imaging (MRI) machine and financial constraints were the reasons for not doing an MRI, which would support the Doppler ultrasound findings. A definitive diagnosis with genetic testing was also impossible due to financial constraints. A diagnosis of KTS was made, and common complications of the disease were excluded. Adequate information and counseling about KTS were relayed to the parents, and the patient was not on any medication. A continuous six-month appointment for the patient's follow-up has been established in the last two years, where complications are sought and adequately treated.

Discussion

Klippel-Trenaunay syndrome is considered sporadic; however, an angiogenic growth factor 1 gene (AGGF1) mutation has been reported (3,8). The isolation of a gene mutation along the somatic lines involving the phosphoinositide 3-kinase pathway, the PIK3CA gene, was also implicated in KTS and other conditions with limb overgrowth (9,10). This enzyme intervenes in the in-utero embryonic cellular growth, and diseases involved in this pathway are classified as PIK3CA-related overgrowth syndrome (PROS) members (9,10).

Patients with KTS majorly present with a triad of port wine stains, vascular malformations, and varicose veins (3). The port-wine stains are abnormal ectatic thin-walled capillaries in the papillary dermis (11). In KTS, the usual venous malformations are the persistence of an embryonic vein (lateral vein) and valveless truncal varicose veins, and their abnormally dilated lymphatics do not connect to the normal lymphatics (11). It usually has a benign course; however, abnormal visceral vasculature resulting in complications may exist and can be life-threatening (11). In this case, hyperpigmentation on the patient's torso and lower limb, with hypertrophy of the right lower limb bones and skin with bone length differences and ectatic veins, are findings in conformity with many reported cases (3,11). KTS must be recognized and managed in children with capillary malformations and limb hypertrophy. Usually, a color Doppler ultrasonography is the initial step for assessing vascular lesions; however, MRI and MR venography can be more useful in determining the extent of underlying malformations. There is no definitive treatment for KTS, only symptomatic medical management with a few needing surgical intervention. However, management that will relieve symptoms, prevent complications, and improve life quality, such as laser therapy for

hemangiomas, compression stockings for varicosity, and corrective orthopedic braces, should be instituted.

Conclusion

Klippel-Trenaunay Syndrome is a rare congenital vascular malformation scarcely reported in our environment. It can be easily misdiagnosed, and treatment and prevention of complications can be delayed.

Declaration

Ethical consideration: Ethical approval was obtained from the Health Research Ethics Committee (HREC) of the Olabisi Onabanjo University Teaching Hospital, Sagamu, Nigeria (reference number OOUTH/ HREC/813/2024AP). The participant and the parents gave assent and written informed consent, respectively.

Authors contributions:

AOOA made substantial contributions to the work's conception and design. She was involved in image acquisition, analysis, and manuscript interpretation. She also substantively drafted and revised the work and contributed to its writing.

AOA contributed to the conception and design of the work, acquiring, analyzing, and interpreting the data and images, substantively revising the work, and writing the manuscript.

SOA contributed immensely to writing, acquiring, interpreting, and substantively revising the data and the manuscript's writing.

ACE contributed to the concept, writing, acquiring, interpreting the data, and substantively revising the work. He also contributed immensely to the manuscript's writing.

ALA contributed to the concept, writing, acquiring, interpreting, and substantively revising the data, and immensely to the manuscript's writing.

AAO contributed immensely to writing, acquiring, interpreting, and substantively revising the data and the manuscript's writing.

All authors read and approved the final manuscript.

Competing interests: We declare no conflict of interest in the manuscript.

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