CASE REPORT

SPECTRUM OF IMAGING FINDINGS IN KLIPPEL-TRENAUNAY SYNDROME AFFECTING LOWER LIMB: A CASE REPORT

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ABSTRACT

Klippel-Trenaunay syndrome (KTS) is a rare congenital disorder featuring triad of vascular malformation, capillary malformations (port-wine stains), and limb hypertrophy. It usually manifests unilaterally in lower limbs. We discuss a case of a 2-year-old boy born with swelling of right lower limb with enlarged right leg. Physical examination showed a grossly enlarged right thigh. There were multiple venous varicosities along with diffuse port-wine stains on the right leg along anterior and lateral aspect. Doppler ultrasound revealed several venous varicosities. Plain radiographs confirmed limb hypertrophy. On the right lower limb MRI, multiple tortuous superficial veins were seen diffusely affecting the skin and subcutaneous tissues with circumferential soft tissue hypertrophy. This massive venous and lymphatic malformation was seen extended inferiorly to the dorsum of the foot and superiorly to the hip joint. These radiological findings were concerning for the diagnosis of Klippel-TrØnaunay syndrome. Medical imaging is the cornerstone in the diagnosis and assessment of severity and complications, follow-up and differentiation of KTS from other similar conditions

Keywords: Klippel-TrØnaunay syndrome; port-wine stain; vascular malformations; venous varicosity

Introduction

The rare and complicated congenital condition known as Klippel-Trenaunay syndrome (KTS) is characterized by limb overgrowth, venous and/or lymphatic abnormalities, and capillary anomalies.[1] PIK3CA-related overgrowth spectrum (PROS) illnesses include KTS, which is thought to be caused by somatic mutations in the PIK3CA gene.[2,3] The triad findings of cutaneous port-wine stain, venous varicosities, and bone and soft-tissue hypertrophy are used to diagnose KTS; lymphatic abnormalities may also be present. Typically, it shows up as unilateral lower limb involvement, although it can also evolve into upper limb and truncal involvement. Spinal structures and visceral organs are seldom affected by venous abnormalities.[4] The disorder's severity can differ greatly, ranging from only cosmetic deformity to severe impairment, thromboembolic events, and chronic pain syndrome. A patient with KTS may exhibit genitourinary and intestinal problems along with acute or persistent bleeding. Low-flow vascular abnormalities are often confirmed by Doppler US, MRI and/or MR angiography, or CT angiography, which is typically used to support clinical diagnosis.[4]

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Case Presentation

We discuss a case of 2-year-old child who presented with unilateral enlarged right leg with edematous and swollen right lower limb. Upon physical inspection, the right thigh was noticeably enlarged. Multiple compressible varicosities, capillary malformations manifested a diffuse port-wine stains along the anterior and lateral side, and limb-length discrepancy were observed in the right leg. Patient was initially investigated with Doppler ultrasonography that identified venous malformation manifesting multiple venous varicosities. Plain radiography verified limb hypertrophy showing bone elongation contributing to leg length discrepancy and adjacent soft tissue thickening. Patient was further advised with MRI right lower limb that clearly depicted venous malformation that were shown as numerous tortuous superficial veins diffusely involving the entire subcutaneous and muscular compartment of right lower limb without respecting facial planes, involving skin and subcutaneous tissues with circumferential soft tissue hypertrophy This whole extensive malformation was extending superiorly uptil hip joint and inferiorly till dorsum of foot. Multiple flow voids were also noted in the lesions showing high flow component. Marrow signal of visualized bones was normal with no evidence of bone bruise. Extensive subcutaneous lymphatic malformation also appreciated involving right lower limb.Based on clinical and radiological findings, diagnosis of Klippel-Trenaunay syndrome (KTS) was made.

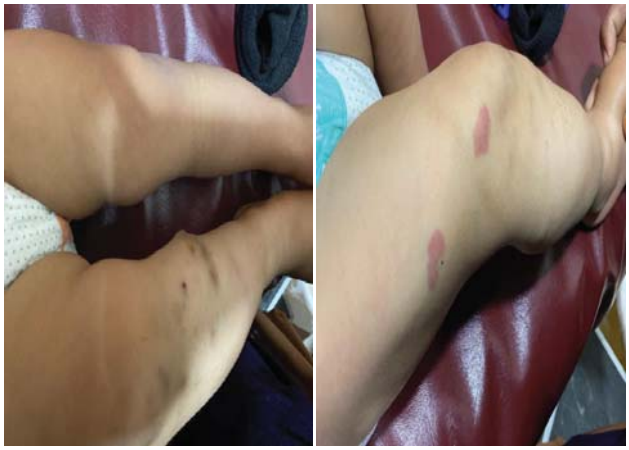


Figure 1a&b: Shows right lower limb hypertrophy with red spots on the skin.

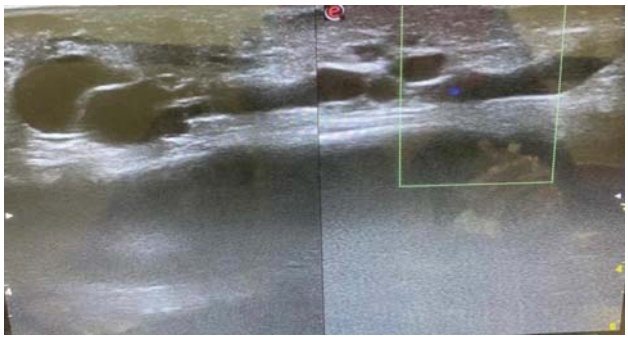
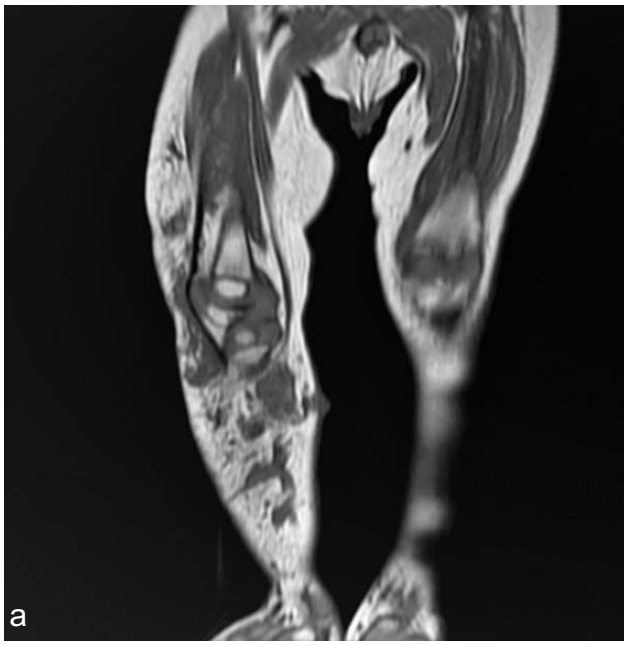
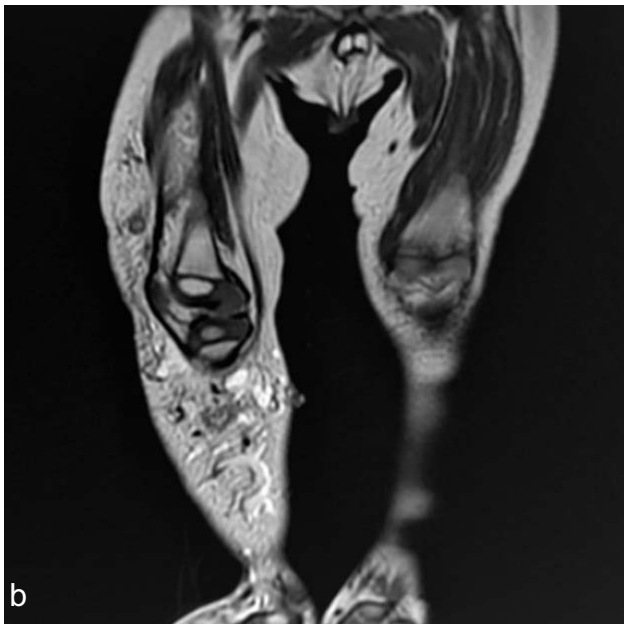
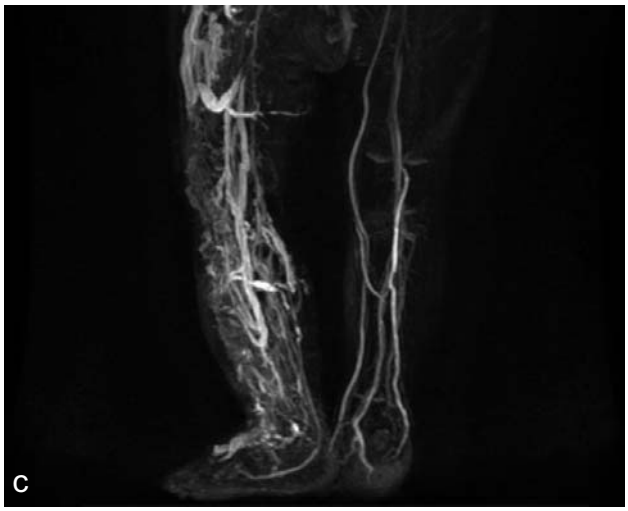


Figure 2: Ultrasound grey scale and colour doppler shows multiple superficial varicosities in right lower limb.







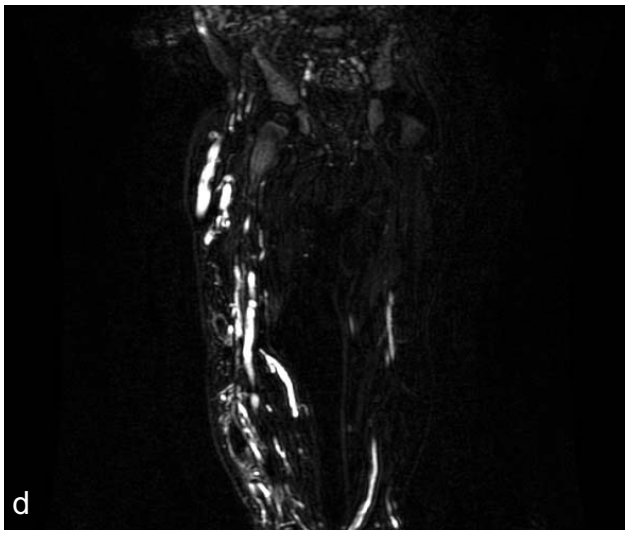


Figure 3a,b,c,d: Shows extensive venous malformation involving right lower limb with limb hypertrophy

Discussion

Rare case, Klippel-Trenaunay syndrome (KTS) triad includes limb hypertrophy, venous varicosities, and vascular malformation. KTS is an uncommon condition with an estimated incidence of 3-5/100,000.[5] Parkes Weber syndrome, Maffucci syndrome, Beckwith-Wiedemann syndrome, and macrodystrophia lipomatosis are significant differential diagnoses for this disorder.[6] The most frequent findings are superficial venous malformations of the unilateral lower limbs or varicosities in unusual distribution, but there have also been reports of deeper extension into the underlying muscle, bone, or even visceral organs such as the liver, spleen, pleura, colon, or bladder (submucosal varices).[6] It has also been possible to identify anomalies of the deep veins, including aplasia, duplications, and aneurysmal dilatation.[6] Another characteristic suggestive of KTS is the persistence of embryonic veins, such as the sciatic vein and the lateral border vein of the servelle.[7] Even though traditional venography is a gold standard method, more noninvasive modalities like color Doppler, CT, or MR venography have largely superseded it because of its intrusive nature. The imaging method most often used to diagnose KTS is color Doppler sonography. Deep venous abnormalities and varicosities in an uncommon place (such as the lateral part of the leg) are strongly suggestive of Klippel-Trenaunay syndrome. Malformations of the vascular (venous) system manifest as a network of circulatory channels with calcified phleboliths and soft tissue components scattered throughout. The low flow vascular malformations show either no flow or monophasic flow on spectral Doppler. The hallmark of this condition is the lack of high flow arteriovenous malformations.[8] A thorough preoperative mapping of the venous system using an additional CT or MR venography may help determine the degree of a deep-seated abnormality and, as a result, may help stop symptoms from getting worse after treatment for superficial varicosities. Since adolescents and young adults make up the bulk of afflicted patients, MRI is a more appropriate primary imaging modality due to the lack of hazardous radiation. CT venography has a substantial process time reduction and a greater spatial resolution compared to MR venography. KTS patients often require lifetime follow-up and are treated conservatively. The major goals of treatment are to control any bleeding episodes, relieve symptoms, and avoid and cure consequences such cellulitis, deep vein thrombosis, chronic coagulopathy, and congestive heart failure.[9]

Conclusion

To sum up, medical imaging is essential for diagnosis, severity and complication evaluation, follow-up, and KTS distinction. When evaluating patients with KTS, plain radiography, phlebography, USG, CT,

MRI, and DSA all have important and complimentary roles to play.

Conflict of Interest: None

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