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Case Report

Radiology Section

Klippel-Trenaunay Syndrome of the Extremities- A Report of Two Cases

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ABSTRACT

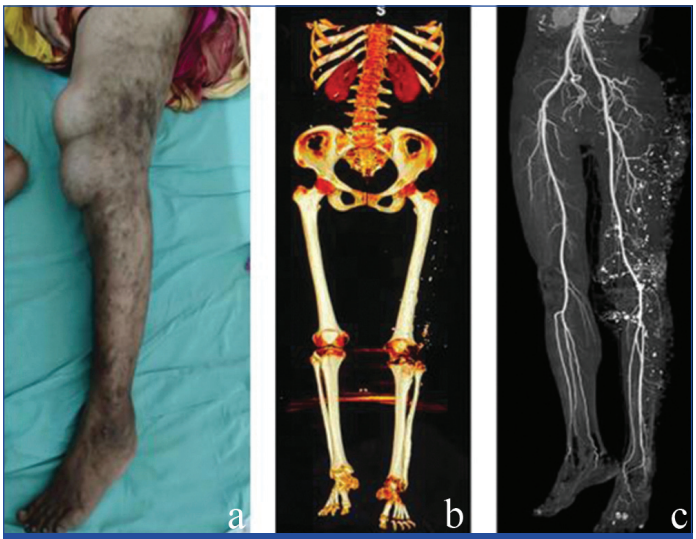
Klippel-Trenaunay Syndrome (KTS) is a rare disorder, with an incidence of one in 100,000 people worldwide. The hallmark features of the KTS include venous malformations, capillary malformations and soft tissue hypertrophy. There is no gender or racial predilection and it manifests in childhood. It usually involves one limb, predominantly the lower limb. But, in rare incidences, upper limb involvement is also reported. The common presentation of KTS is pain with spontaneous cutaneous haemorrhage, soft tissue enlargement, venous thrombosis, leg length discrepancy, cortical thickening, macular patches and phleboliths. The present case report described two patients (both females) with KTS, who visited the hospital. One of them was an adult female, presented with left lower limb swelling. Computed Tomography (CT) showed, multiple dilated lower limb veins, draining into the internal iliac and deep femoral veins. Following this, the patient had undergone one cycle of Ultrasound Guided Sclerotherapy (UGS) and was advised to follow-up. The patient underwent subsequent two cycles of UGS at an interval of three months. Another patient was of the paediatric age group, presented with swelling and pain in the left lower limb. Magnetic Resonance Imaging (MRI) revealed, multiple veno-capillary malformations with a Persistent Sciatic Vein (PSV). Liposuction of the bilateral gluteal region was performed with the application of topical Neosporin and was advised to follow-up. After five months, Ultrasonography (USG) of the bilateral gluteal region was done, which was normal with no obvious residual disease.

Keywords: Capillary malformations, Cutaneous lesions, Dilated veins, Venous malformations

CASE REPORT

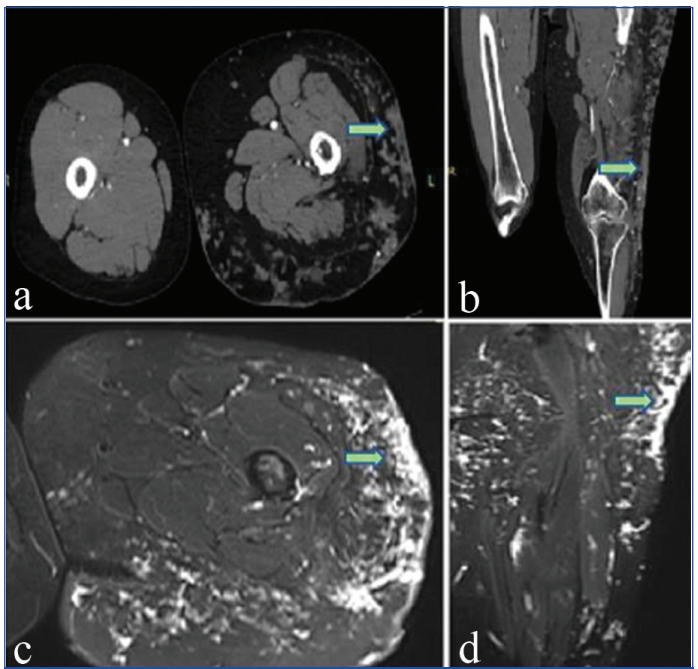
Case 1

A 30-year-old woman, presented to the Department of Radiology, with the chief complaint of an increase in swelling and pain in the left lower limb for the past two weeks. The patient had similar complaints for the past 25 years. There was a history of visiting another hospital in Kolkata and was diagnosed to be having KTS. But was not clear about the treatment taken and there were no documents. On examination, the patient had a macular patch on the left lateral thigh with a few varicose veins and soft tissue swelling [Table/Fig-1].



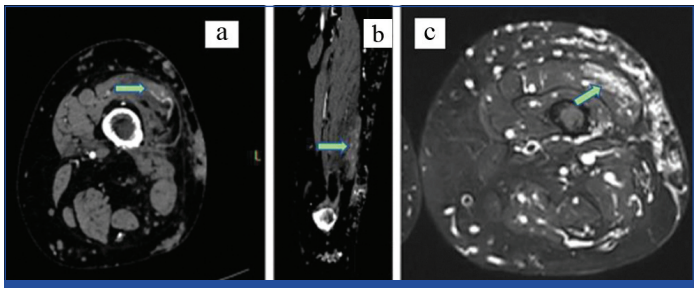
[Table/Fig-1]: a) Macular patch over the lateral aspect of the left thigh b) Shows left limb lenghtening and cortical irregularity c) Shows multiple calcifications.

Doppler Ultrasonography (DU) of the left lower limb showed, multiple dilated tortuous veins with dilated perforators throughout the left lower limb, more on the lateral aspect. CT angiography was performed, which revealed, multiple subcutaneous soft tissue lesions in the posterior and medial aspects showing more enhancement in the venous phase than in the arterial phase [Table/Fig-2]. One such lesion in the vastus lateralis muscle measured 2.6×13.2 cm with the feeding artery from the distal superficial femoral artery [Table/Fig-3]. In addition to this, there were multiple dilated tortuous subcutaneous and muscular veins of the left lower limb (Lateral>medial), extending from the foot to the gluteal region, draining into the left internal iliac and the deep femoral vein. Common iliac, internal iliac and deep femoral veins were dilated, as compared to the right side [Table/Fig-4]. Diffuse hypertrophy of the subcutaneous fat was noted in the suprapatellar bursa and Hoffa's fat pad and there was muscular atrophy of the entire left

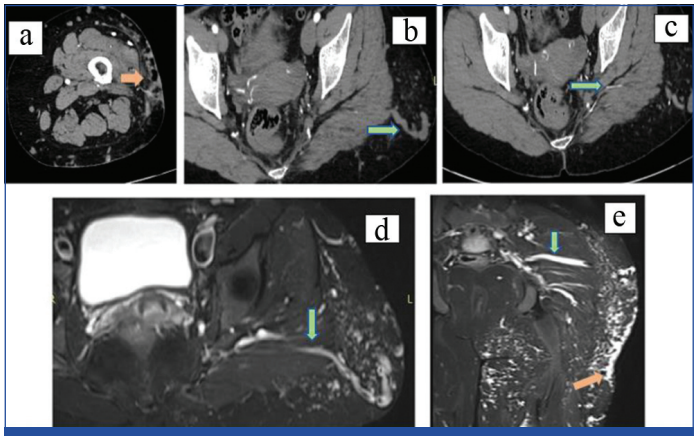


[Table/Fig-2]: a) CT axial, b) CT coronal, c) MRI, Short Tau Inversion Recovery (STIR) axial, d) MRI, STIR coronal. Images showing subcutaneous lesions (green arrows).

lower limb. There were subcutaneous and intermuscular grouped and discrete calcifications. Other features like a mild increase in the vertical length of the femur, tibia and fibula were observed with cortical irregularity and periosteal thickening [Table/Fig-1]. Further confirmation of the features was done with MRI. T1, T2, and STIR sequences. The patient had undergone one cycle of USG-guided sclerotherapy and was advised to follow-up for the next cycles. The patient had subsequent two cycles of UGS with an interval of three months between each cycle. Further, the patient did not visit the hospital after the third cycle, no follow-up was done.



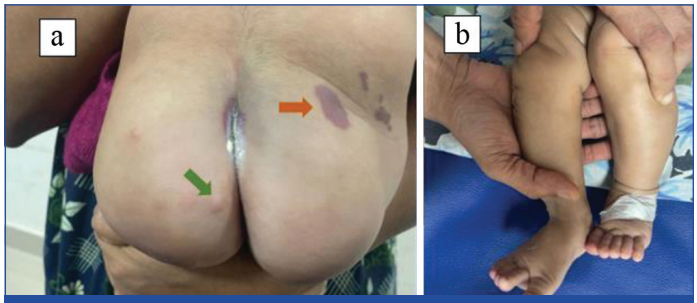
[Table/Fig-3]: CT Axial (a), Coronal (b) and MRI STIR axial (c) Shows an intramuscular haemangioma with a feeding artery (green arrows).



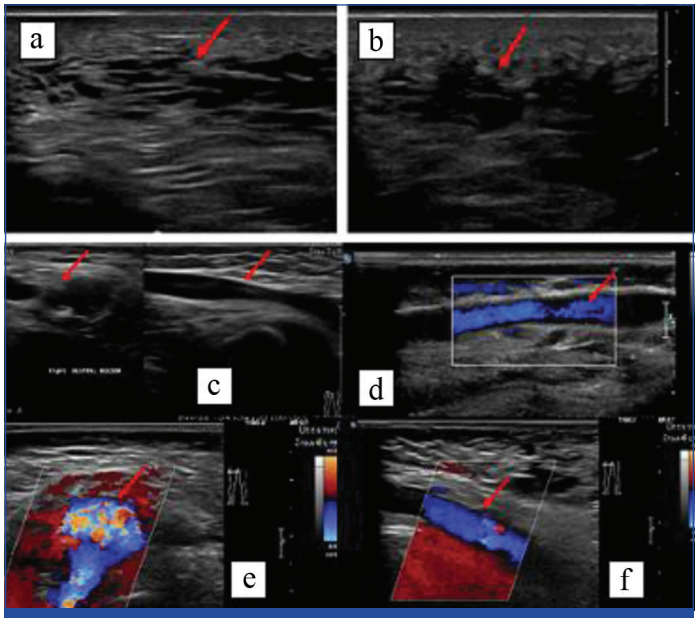
[Table/Fig-4]: Axial CT images (a,b,c) d) MRI STIR axial e) Coronal. Images shows subcutaneous dilated veins (orange arrows) draining into the internal iliac vein (green arrows).

Case 2

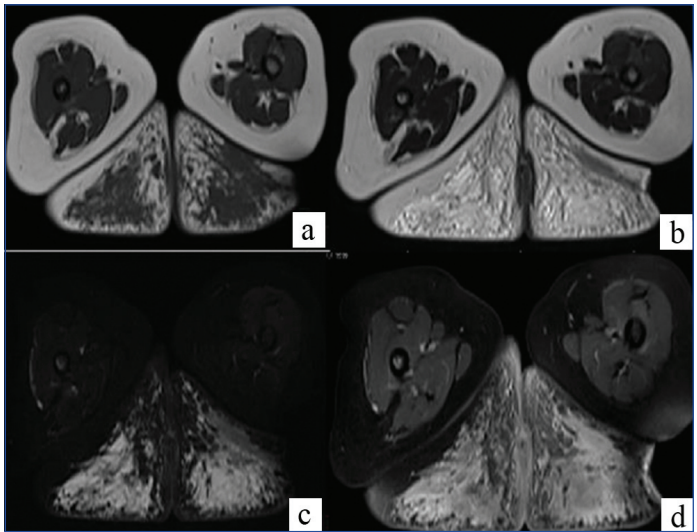
A four-month-old female child, who was born out of a non consanguineous marriage presented to the Department of Radiology, with the chief complaint of swelling along the lower part of the left gluteal region, which was first noticed at the age of two months. One month ago, the child developed pain with an increase in size of the right buttock, swelling, and redness over the right lateral malleolus, for the past four days. On examination, there was a diffuse swelling of the bilateral gluteal region with a port-wine naevus on the left buttock [Table/Fig-5]. Ultrasound of the bilateral gluteal regions showed thickening of subcutaneous soft tissue with multiple cystic spaces, slow flow vascular channels, and intermuscular plane tubular anechoic structures with thin internal septations and internal echoes [Table/Fig-6]. Contrast MRI pelvis showed hypertrophied soft tissue of the skin and subcutaneous plane in the bilateral gluteal region, foot and toes. These areas showed, heterogenous enhancement with cystic areas and intervening septae, appearing like a vascular malformation with the predominantly lymphatic component [Table/Fig-7]. These areas were seen extending anteriorly, through the intermuscular plane into the presacral region and pelvis, where, the cystic components appeared encasing the rectum and bilateral iliac vessels. These were extending through the sciatic foramen on both sides (right>left) into the intermuscular plane of the gluteal region, sacral canal, extending along the sacral nerve roots to the right ischiopubic ramus, bilateral labial majora (right>left), and perineum. The PSV was present, which was seen as a tortuous tubular structure along the right sciatic nerve draining into the internal iliac vein [Table/Fig-8]. The lower limb veins appeared dilated and tortuous. Liposuction of the bilateral gluteal region was performed with the application of topical Neosporin and was advised to follow-up. After five months, USG of the bilateral gluteal region was done, which was normal with no obvious residual disease.



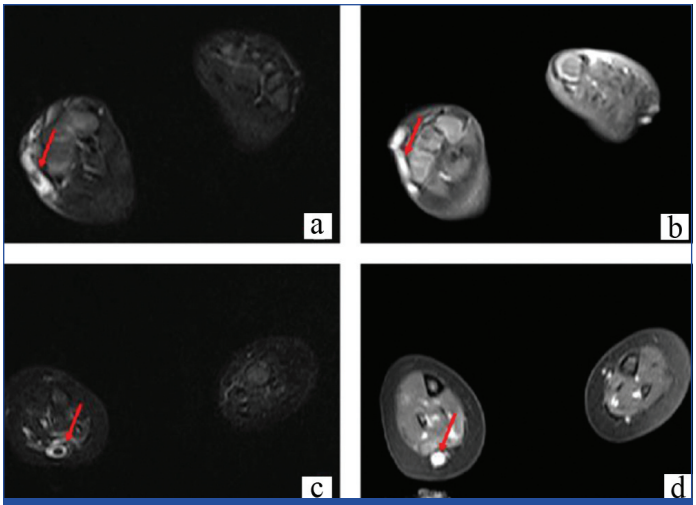
[Table/Fig-5]: (a) Shows port-wine nevus (orange arrow) in the left gluteal region with bilateral gluteal swellings (green arrow). (b) Shows mild swelling of bilateral limbs.



[Table/Fig-6]: (a) and (b) USG of the gluteal region shows thickening of subcutaneous plane with multiple microcystic lesions (red arrows). (c) Shows tortuous vein in the intermuscular plane (red arrows). (d-f) Colour doppler: shows the same tortuous vein with colour uptake in right gluteal region extending into thigh, likely extending along the course of sciatic nerve (red arrows).



[Table/Fig-7]: MRI T1W (a), T2W (b), T2W Fat-saturation (Fat-Sat) (c) Post contrast T1W Fat-Sat (d) Axial images show hypertrophy of the subcutaneous soft tissue of bilateral gluteal region with microcystic areas and heterogenous post-contrast enhancement lymphatic malformation likely microcystic lymphangioma.



[Table/Fig-8]: MRI: T2W Fat-Sat (a and c) Post contrast T1W Fat-Sat axial (b and d) Bilateral lower limbs respectively, showing dilated tortuous Right lateral marginal vein (a and b) (red arrow) Right short saphenous vein (c and d) (red arrow).

DISCUSSION

The KTS was discovered by two physicians in 1900, Klippel and Trenaunay after which it was named [1,2]. Defects in the angiogenetic protein and VG5Q genes are one of the theories [3]. The failure of regression of the embryonic veins causes venous stasis and impaired venous return, which in turn can lead to tissue overgrowth. Hypertrophy is a classical feature of KTS. Capillary malformations are common with cutaneous manifestations. Patients also present with varicose veins involving both the superficial and the deep veins, usually on the lateral aspect, an unusual site for varicosities [1,4,5]. The commonly draining veins of the varicosities are the deep femoral vein and internal iliac vein. Superficial veins involvement is also seen. Persistent Embryonic Veins (PEVs) are characteristic of KTS. The Lateral Marginal Vein (LMV), which is a part of the superficial venous system and the PSV, part of the deep venous system is more commonly involved and these both usually regress before birth [3]. Complications include dermatitis, thrombophlebitis, coagulopathy, pulmonary thromboembolism, and congestive cardiac failure. Associations of KTS are polydactyly, syndactyly, congenital hip dysplasia, vascular malformations of the colon, rectum and urinary bladder, spinal, liver, and splenic haemangiomas, and lymphangioma of the limb [1]. Genetic testing like chromosome study on G banded chromosomes from peripheral blood cultures can now be implemented, which helps in the identification of the angiogenesis factor for further therapeutic intervention [2,6]. The treatment of KTS included surgical and conservative management with surgery and debulking being the treatment of choice, however, series of cycles of sclerotherapy and radiofrequency ablation in younger patients. The early detection of this syndrome, can help in the early treatment and prevent the transmission to successive generations is currently at the forefront in the genetic field [6].

In a study conducted by Huang FL et al., a two-year-old child had presented with diffuse hypertrophy of the right upper limb with port-wine neavus and was suggested MRI with Magnetic Resonance Angiography (MRA) which showed, soft tissue hypertrophy of the right upper limb, with multiple capillary, lymphatic and venous malformations, which were similar to the findings found in the present case report. The child also had limb discrepancy with cortical erosions which were seen in the present case report as well. The child had undergone amputation of the right arm as the treatment of choice [6]. Similarly, a study conducted by Kharat AT et al., a six-year-old child, presented with painful port-wine stain on the lateral aspect of the right thigh, following which MRI revealed multiple irregular tortuous subcutaneous venous channels, with soft tissue hypertrophy of the right limb [7]. There was increase in the length of the right femur causing abnormal gait. These findings were similar to present case. However, the unique findings in the present case included the presence of an intramuscular haemangiomas, cutaneous lesions, lymphangiomas, dilated venous channels draining into PEVs. Arasu A et al,, in their study on 13-year-old female with KTS, showed, findings which were similar to the index case, like soft tissue hypertrophy, limb lengthening with bony changes, subcutaneous lesions, intramuscular haemangiomas, dilated tortuous venous channels and phleboliths [8]. The unique finding in their study, was an associated splenic haemangioma.

Karmacharya RM et al., in their case series on seven patients with KTS states that, all the patients had typical cutaneous vascular stains, venous malformations with three of the patients having PEVs, and one of them developing multiple ulcers as complication [9]. Plain radiographs and Multiplanar Reconstruction (MPR) CT images, help to determine the limb hypertrophy and to screen for limb-length discrepancies as a discrepancy of less than 2 cm does not require any treatment. MRI is both, sensitive and specific for vascular anomalies and overgrowth and is regarded as the imaging of choice for evaluating most of the findings in KTS. It also helps in the differentiation of soft tissue and the addition of intravenous gadolinium contrast, can help discriminate the venous malformations of KTS from the arterial involvement of Parkes Weber Syndrome (PWS). Use of CT scan and CT angiography is limited, due to the radiation risk and the potential renal toxicity of contrast usage especially when MRI is readily available and can better delineate vascular anomalies. Contrast Enhanced Computed Tomography (CECT) scan may be particularly useful in guiding management planning prior to contemplated interventional procedures. Venography may also assist in planning, prior to interventions and can even be performed during the actual interventional procedure [9,10].

In the patients of the present case report, CECT and MRI helped in identifying the vascular malformations, subcutaneous hypertrophy, bone cortical thickening of the bone, and dilated venous channels that lead to the confirmation of the KTS.

CONCLUSION(S)

The KTS is a syndrome, that usually occurs in the paediatric age group and early diagnosis of this syndrome can lead to tailored medical and surgical care depending on the extent of the disease involvement. In the present case report, the authors have found that, the dilated veins were seen draining into the internal iliac and deep femoral vein in the elderly patient. In the paediatric patient, the authors have found that the earlier diagnosis of the disease had led to the complete cure after the surgery. MRI with its key findings plays a vital role in the diagnosis.

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