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Femur Fracture in Klippel Trenaunay Syndrome: A Case Report

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Learning Point of the Article:

This case reports highlights the importance of and difficulties in planning as well as management of a long bone fracture in a case with associated rare vascular malformation such as Klippel-Trenaunay Syndrome in the affected extremity.

Abstract

Introduction: Klippel-Trenaunay Syndrome (KTS) is a medical condition that involves the abnormal enlargement of bones and soft tissues, along with the presence of arterio-venous malformations and cutaneous vascular nevus. This is an uncommon condition that is present from birth and affects the blood vessels. There are relatively few documented cases of how to treat fractures in the lower limbs of individuals with this condition.

Case Report: A 37-year-old Indian male patient presented to our emergency room with a history of trivial trauma resulting from slipping and falling on the floor. From a clinical perspective, there was a bone deformity observed on the right thigh, accompanied by large dilated veins on the affected limb. The radiographs revealed osteoporotic bone with a fracture in the middle third of the right femur, as well as several small calcified masses dispersed throughout the affected lower leg. A Computed Tomography angiography was performed, revealing the presence of developmental venous malformations along with dilated deep and superficial venous channels. The diagnosis of KTS was ultimately confirmed by integrating clinical and radiological observations. The patient had surgical management with closed intramedullary nailing. Subsequent examinations showed strong indications of bone union without any significant complications.

Conclusion: the surgical treatment of orthopaedic injuries in patients with KTS has been demonstrated to be linked to higher amounts of blood loss during the operation. In such circumstances, it is essential to do a comprehensive clinic-radiological assessment and careful preoperative planning to effectively treat or prevent surgical problems in these patients.

Keywords: Fracture, Femur, Klippel-Trenaunay Syndrome

Introduction

Klippel-Trenaunay syndrome (KTS) is a rare congenital condition characterized by a triad of (a) cutaneous naevi (capillary malformations- the "port wine stain"); (b) varicose veins or venous malformations; and (c) hypertrophy of bones and soft tissues of the extremity. [1] 70% of cases are present in the lower limb and 11% in the upper limb. It is one of at least nine vascular malformation syndromes that can affect the limbs. The condition commonly presents itself with pain in the lower limb, bleeding, functional limitation, and cosmetic sequelae as a result of venous malformation or tissue over growth. [2]

Although the aetiology is unknown, there are multiple proposed theories such as sympathetic ganglion defect, deep vein disorders, embryonic mesodermal anomalies causing persistent arteriovenous malformations, mixed meso-ectodermal dysplasia and genomic mutations. [2]

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Author's Photo Gallery


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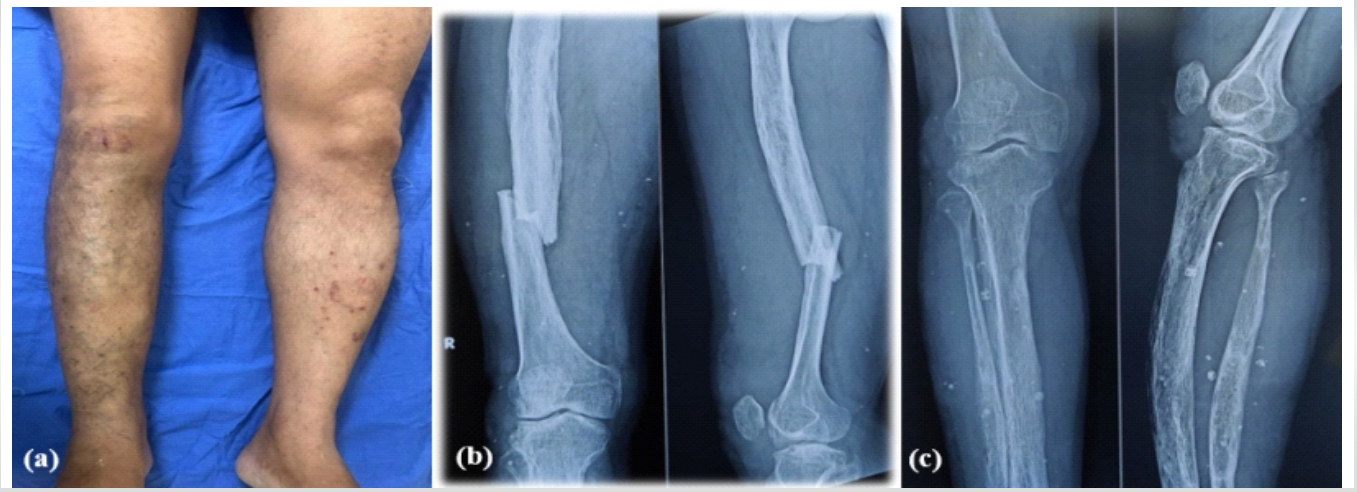

Figure 1: Clinical and Radiological presentation of the patient. [a] clinical picture of the patient's leg on presentation showing multiple enlarged venous engorgement, [b] plain radiographs showing fracture of the middle shaft of the femur, uneven cortical thickness and osteoporotic bone with multiple phleboliths scattered over the affected limb. [c] plain radiograph showing anterior bowing of the tibia with loss of uniform cortical thickness, evidence of osteoporosis and multiple, scattered phleboliths.

If operating on the limb, the pathology of this condition can prove a very challenging management scenario. Venous malformations have been reported to bleed excessively during orthopedic procedures, requiring copious blood transfusions. With regard to orthopedic operations, it is also known that patients with KTS can have an osteoporotic bone. This can prove difficult when making decisions regarding the most appropriate management of any elective procedure or traumatic bony injury. [3]

There have been a limited number of previously documented cases of femur fractures in this challenging group of patients. Hence, it is necessary to gather additional experience and information on such cases to establish a comprehensive, evidence-based strategy for management.

Case Report

A 37-year-old male patient arrived at the emergency room with complaints of intense pain in his right lower limb after a minor accident of slipping and falling at home. The clinical assessment revealed a deformity and bony protrusion in the middle one-third of the right thigh, accompanied by conspicuous engorged veins throughout the right leg. (Fig.1) The patient reported a longstanding history of weakness and inability to fully support their weight on the right lower limb after walking long distances from childhood.

Radiographic findings showed:

1. Transverse fracture of middle third of right femur
2. Asymmetrical diaphyseal cortical thickening
3. Irregular bone margins and osteopenia
4. Narrowing of the medullary canal of the femur, tibia and

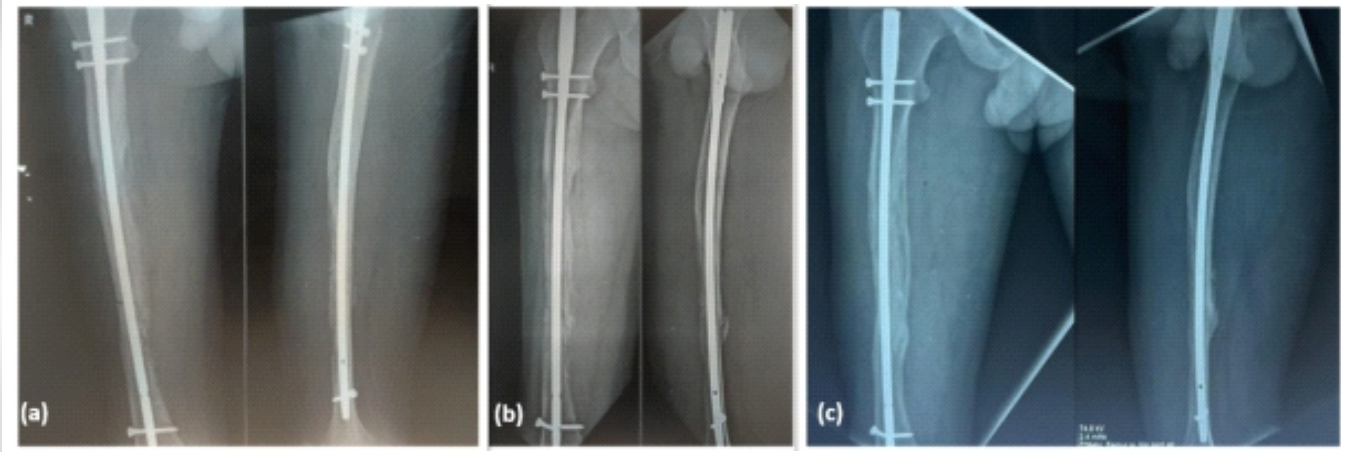

Figure 2: Follow-up radiographs. [a] 2-month follow-up radiograph shows callus formation at the fracture site. [b] 8-month follow-up radiograph shows breakage of the distal screw (asymptomatic), [C] 36-month follow-up radiograph shows complete union of the fractured and osteoporotic femur with non-uniform cortical thickness at the remodeling site.

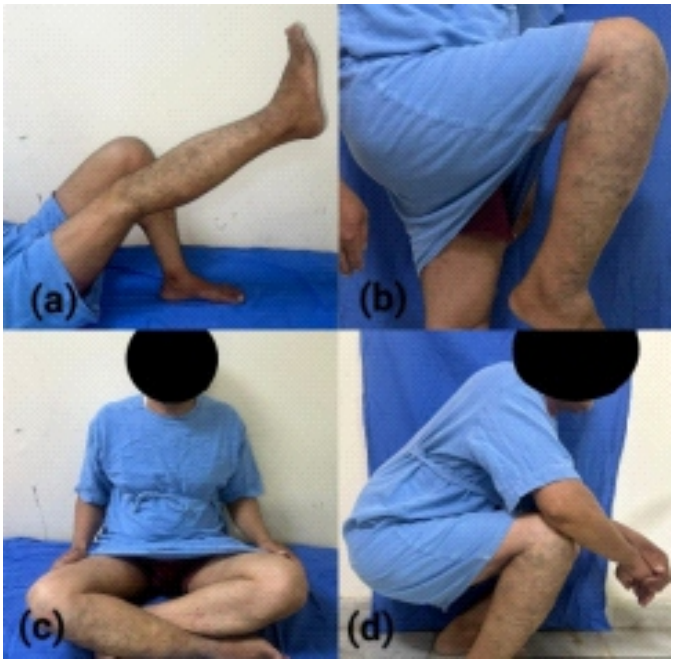

Figure 3: Follow-up of functional outcome. (a and b) shows flexion and extension of the ipsilateral knee after 36 months of surgery. (c and d) show cross-legged sitting and squatting after 36 months of surgery.

fibula

5. Multiple, tiny calcified tissues scattered over the entire limb suggestive of phleboliths. (Fig. 1b)

We suspected the presence of a vascular problem in the right lower limb due to the significantly dilated veins. To confirm this, we scheduled a Computed Tomography Angiography of the right lower limb to eliminate the possibility. The imaging results verified the existence of dispersed phleboliths and revealed the presence of many enlarged clusters of blood vessels, indicating the presence of a developmental venous malformation in the anterolateral thigh with feeding vessels from both superficial and deep veins. The hematological tests revealed a moderate level of anaemia (hemoglobin-7.4g/dL) and a severe level of thrombocytopenia (Platelet count 0.65 lakhs). All other blood values were within the usual range.

Based on clinical findings, radiological and blood investigations, we reached a diagnosis of KTS/ Klippel-Trenaunay-Weber Syndrome/Angio osteo-hypertrophy Syndrome. The classical triad features were consistent with those found in our patient which confirmed the present diagnosis.

The patient was transfused 4 units of Platelets (Single Donor Platelets) and 2 units of whole blood preoperatively to preliminarily recover the obvious intra operative blood loss. By reaching the correct diagnosis at an early stage, we were prepared with our complete surgical armament to perform closed intramedullary (IM) nailing for the fracture. Unreamed nailing was planned to reduce blood losses from serial reaming of the bone. 1g Tranexamic acid was administered before the start of surgery. Although, intraoperatively reaming had to be done to prevent cortical disruption which seemed probable while inserting an unreamed nail. There was excessive intraoperative bleeding during the fixation of distal screws owing to the complex vascularity in the lateral aspect of the lower thigh. This required 1 unit of whole blood transfusion intraoperatively for hemodynamic stabilization. The surgery was completed in 126 min. The patient was transfused 1 unit of whole blood postoperatively and was kept in the Intensive Care Unit for 24 h for observation after which he was transferred to the general ward. Partial weight bearing was allowed after 7 days and full weight bearing after 1 month from the index surgery. No post-operative complications were seen and the patient was asked to follow-up at regular intervals. 8-month post-operative radiograph showed breakage of the distal screw which was asymptomatic. The patient was pain-free and was followed up until 3 years after surgery and the check radiograph showed excellent results in terms of functional status, union and remodelling of the fractured femur. (Fig. 2&3).

Discussion

Various underlying etiologies like sympathetic ganglion defects, mixed mesodermal and ectodermal dysplasias, genetic mutations, venous hypertension and various varicosities have been found to be associated with KTS. There has not been any gender or racial preponderance and the incidence has been found to be sporadic with lower limbs being most commonly affected. [2]

A thorough history and clinical examination are usually sufficient to make a diagnosis in these cases but imaging methods such as CT and Venous and Arterial Color Doppler of the affected limb offer confirmation and better preoperative planning as well as follow-up plan.[2]

KTS is not usually life-threatening, and management of the condition is conservative with lifelong follow-up. However, vascular and bony malformations may lead to challenges in managing patients requiring orthopaedic surgery.[4]

This is one of the few reported cases of a femoral fracture in an adult KTS patient, fixed with closed IM nails. Multiple options have been considered when deciding the optimal fracture fixation technique for these patients with varying results.

Conservative management has been attempted, but with poor results mainly due to increased chances of nonunion of the dysplastic femur. External fixation is deemed unsafe due to the patient's fragile skin and risk of bleeding, given the patient's history of bleeding profusely from pin sites. Plating has the

highest risk of intra-operative bleeding due to the extent of the wounds.

Gupta et al. reported a case of femoral fracture in KTS patient with external fixation followed by skeletal traction. Although this patient developed a profound shortening of 5cm. [5]

Patel et al. used pediatric-size Titanium Elastic IM Nail System (TENS) for a diaphyseal fracture of the femur in a 34-year-old KTS patient due to narrow medullary canal of the affected femur. Delayed fracture union was managed by low intensity pulsed ultrasound (LIPUS) and the fracture showed radiological union 2 years after surgery. [6]

Nahas et al. performed open reduction and internal fixation on a patient of KTS with a femoral shaft fracture. They required 15 units of blood intraoperatively due to significant blood losses. The patient developed delayed union and was advised LIPUS for 20 weeks. [7]

Therefore, bleeding from vascular malformations in KTS is a well-reported challenge in the operative management of fractures.

Tsaridis et al. reported the first case of a femoral shaft fracture in a 42-year-old female with KTS fixed with an IM nail and required 10 units of red blood cells (RBCs) during and after surgery.[2] Notarnicola et al. reported management in a 52-year-old male with an IM nail only requiring 4 units of RBCs. Union was achieved in both these cases. [3] Our patient also required 4 units of platelets, 2 units of whole blood preoperatively and 1 unit each of whole blood intra and postoperatively.

Choi et al. also reported a case of 4-months old elsewhere operated femoral shaft fracture with metal failure. The diagnosis of KTS was made after angiography and a two-stage surgery was planned for removal of the implant with embolization of the arteriovenous malformations. IM nailing was done a week later when the patient stabilized. [8]

Yamaguchi et al. planned to manage a case of old femoral neck fracture in a 47-year-old KTS patient with total hip arthroplasty. Although, due to significant intraoperative blood losses (4L approximately), the plan was changed intraoperatively to a resection arthroplasty. [9]

The aforementioned literature review of similar cases of femoral fracture in patients with KTS has shown that the management of these fractures is complex and requires extensive preoperative planning. The timing of surgery plays an important role in determining the possible outcomes and complications. Implant choice and surgical approach proves to be vital to maximize the chances of fracture union.

Extensive bleeding during surgery is a recognized risk in these patients. With no established techniques to eliminate this, nevertheless, efforts should be made to minimize bleeding as much as possible. Although, blood products such as RBCs, Whole Blood, Fresh Frozen Plasma and Donor Platelets should always be kept ready to cater to the needs of the patient for his/her wellbeing and to prevent any hemodynamic instability. Management strategies should be tailored according to fracture pattern, femoral canal diameter and current skin and vasculature within the affected limb. [10]

Conclusion

Managing cases with femoral fracture in patients with KTS is an extremely complex issue and needs careful surgical planning and management. Despite using the minimally invasive approaches, the risk of extensive intraoperative bleeding is always present, requiring blood transfusions both intra and post operatively. Main operative complications include extensive hemorrhage and delayed bone healing. Hyperalgesia and allodynia although rare, may occur post operatively. To conclude, KTS is rare entity and long bone fractures in these cases impose a mammoth challenge for planning and management. The approach to this group of patients must involve a multidisciplinary team of experienced radiologists along with skilled orthopedic and vascular surgeons. Identification of such pathologies very early in management ensures a successful outcome of surgery and patient satisfaction.

Clinical Message

1. Careful clinical and radiological examination is necessary in patients where suspicion of an underlying pathology arises.
2. Vigilant approach and an active management plan should be devised while operating cases of extremity fractures in patients with KTS.
3. Since the potential for large amount of blood loss is always present, blood derivatives like whole blood and platelets should always be kept ready, if needed, for transfusions.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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