INTRODUCTION

Klippel-Trenaunay Syndrome (KTS) is a rare, congenital, vascular disorder affecting one or more limbs. Originally, it was defined as a triad including port wine stain, varicose veins and bony and soft tissue hypertrophy. We present a case of a 20-year-old female who walked with a limp. Because of swelling of right leg she was sent for Doppler study which picked up dilated arteries and increased blood flow velocity. The impression of KTS was further strengthened by unique nuclear medicine and radiological findings.

CASE REPORT

A 20-year-old non-diabetic, normotensive female presented with right leg swelling, skin pigmentation, itching and scaly skin. There was history of limping since childhood.

On examination there was oedema of the right leg, which was more marked over the calf; the skin was discoloured and scaly. Prominent venous varices were visible over the back of thigh. The right leg was bigger with a longer length (82 cm vs. 79 cm), calf girth (30.5 cm vs. 24.5 cm), mid-thigh girth (33.5 cm vs. 29.5 cm). On hematological examination, she was mildly anaemic with haemoglobin of 10.3 gm/gl. The renal function tests, liver enzymes and blood glucose levels were normal. She was sent for a lower limb Doppler study in view of the oedema. There was thickening of the skin and subcutaneous tissue but no lymphoedema was seen. Duplex scan showed dilated right lower limb arterial system. There was marked difference between the diameter of blood vessels of the affected limb as compared to the normal side (Figure 1 A, B and C). Blood flow velocities were almost twice on the right side as compared to the left side with a low resistance pattern and a high diastolic component. There was spectral broadening due to turbulence. X-rays of lower limbs showed distinct osteohypertrophy of right femur as compared to the left femur (Figure 2).

A three phase bone scan was done which revealed a markedly increased flow, expanded blood pool and raised bony uptake. All ratios between the two limbs (flow, blood pool and uptake) were around 8 times higher on the right side, confirming the hypertensive nature of the pathology (Figure 3 A and B). Patient was managed conservatively with compression stockings and advised to have left heel inserts (on the normal side) to compensate for unequal leg growth. Infection healed on conservative management. Follow-up over 2 months has not shown infection recurrence. Patient is advised follow-up every 6 month.
DISCUSSION

The etiology of KTS is unknown. One theory is that KTS may be caused by a mesodermal defect during fetal development causing microscopic arterio-venous communications. Another theory suggested intrauterine damage to the sympathetic ganglia or inferomedial tract leading to microscopic A-V communications. Most cases are sporadic, although a few cases have shown an autosomal dominant pattern of inheritance. In Pakistan Klippel-Trenaunay syndrome has been reported in children under 10 years of age with involvement of lower limb, upper limb, nose, tongue and jaws. All patients manifested the typical features of KTS i.e. hemangiomas and port wine staining with bony and soft tissue hypertrophy.

In a series of 252 patients of KTS, 63% patients had all three features and 37% had two out of three features. Varicosities were present in 72% and limb hypertrophy in 67% of the patients. It has been reported that the proportion of patients lacking a port wine stain can reach even 68%. KTS can be associated with ulcers, hyperhidrosis, and pain in the affected extremity. There can be osteoporosis and compensatory scoliosis as a complication of the differences in leg length.

In this case the disease led to right leg enlargement and elongation with occasional skin infection. In many instances, a thorough history and physical examination are all that are needed for diagnosis but a high index of suspicion is needed and the diagnosis can be missed easily. This case remained undiagnosed for almost 20 years despite several specialist medical consultations. The presence of limb oedema led to the suspicion of deep vein thrombosis and the patient was sent for a Doppler study to rule out this possibility. The presence of enlarged vessels and varices led to an initial suspicion of KTS which was reinforced by nuclear medicine and X-ray studies. Of special interest is the fact that nuclear medicine has not been reported in the diagnosis of KTS despite the elegant demonstration of the vascular nature with enhanced metabolic bone activity of the disease on a three phase bone scan.

The differential diagnosis of a hypervascular limb with expanded blood pool and increased bony uptake would include Paget's disease, osteomyelitis, primary bone tumors and other vascular lesions e.g. haemangioma giving a similar appearance in the three phase bone scan but the clinical presentation would be different (Table 1).
In Paget’s disease there is blooming appearance of areas of intense but focal uptake; clinically there is bony dilatation. The pelvis is commonly involved followed by the spine, skull, femur, scapula, tibia and humerus. In osteomyelitis, there is arterial hyperemia with progressive focal skeletal uptake with relative soft tissue clearance in (delayed) third phase of bone scan. Clinically, the acute onset, signs of infection and pain lead to correct diagnosis. In children, there may be a cold area seen if osteomyelitis is associated with infarction. In cellulites there is (delayed) venous hyperemia and persistent soft tissue activity. There is no skeletal uptake on delayed images. In septic bone disease (mostly involves joints) there is increased focal uptake in all three phases of bone scan. The clinical setting is dramatic with pain, fever and redness. In benign and malignant bone tumours, the general pattern is focal increased uptake of the involved areas. In blast crisis there may be a diffusely increased uptake that is greater at the ends of long bones. Combined with an X-ray and Doppler scanning, the condition can be very accurately investigated and followed-up. Currently, laser treatment can be effective in lightening the colour of port-wine stain.

Surgical intervention in the treatment of varicosities and venous malformation is controversial. Venous stripping, ligation, excision, or sclerotherapy are contraindicated unless it involves a superficial system with normal or mild reflux of deep venous system. Some 90% of treated varicosities will redevelop; treatment can provide lasting improvement only for a few years. For greater limb discrepancies, orthopaedic surgery may be considered. Possible procedures include osteotomy, epiphysiodesis, or epiphyseal stapling. Follow-up patients with KTS should be monitored at least annually and more often if indicated. KTS is not always a static disease process. If progression of disease arises, imaging studies should be undertaken. Medical and/or surgical intervention should be done if indicated.

This case highlights the findings on clinical examination, Doppler ultrasound, X-ray and three phase bone scan. The combination of plain X-ray with either Doppler or three phase bone scan would have given the necessary information to confirm the suspicion and this algorithm might be useful in diagnosing limb asymmetries.

**REFERENCES**


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**Table 1:** Comparisons of three phase bone scan and clinical findings.

<table>
<thead>
<tr>
<th>Disease</th>
<th>Flow</th>
<th>Blood pool</th>
<th>Static (delayed)</th>
<th>Clinical findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pagets disease</td>
<td>+</td>
<td>+</td>
<td>Focal uptake</td>
<td>Bony swelling, distortion, bowing</td>
</tr>
<tr>
<td>Acute osteomyelitis</td>
<td>++</td>
<td>++</td>
<td>Focal uptake</td>
<td>Pain, redness, fever</td>
</tr>
<tr>
<td>Chronic osteomyelitis</td>
<td>+</td>
<td>+</td>
<td>Focal uptake</td>
<td>Pain, redness, fever</td>
</tr>
<tr>
<td>Primary bone tumours</td>
<td>_</td>
<td>_</td>
<td>Focal uptake</td>
<td>No fever but bony swelling present</td>
</tr>
<tr>
<td>Cellulitis</td>
<td>++</td>
<td>++</td>
<td>_</td>
<td>Redness, fever, pain</td>
</tr>
<tr>
<td>Klippel-Trenaunay syndrome</td>
<td>+++</td>
<td>+++</td>
<td>Diffuse uptake</td>
<td>No signs of inflammation</td>
</tr>
</tbody>
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Klippel-Trenaunay syndrome