Unusual Age at Presentation of Klippel-Trenaunay Syndrome

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ABSTRACT

Klippel-Trenaunay (KT) syndrome is a congenital vascular disorder which is rare and affects one or both limbs. It has incidence of about 2-5 in 100,000. French physicians Klippel and Trenaunay first described this syndrome in the year 1900. They named the syndrome as "nevus vasculosus osteohypertrophicus". In 1907, Park Weber named the same condition hemangiectatic hypertrophy. The etiology is unknown. Though it is a sporadic condition, paradominant inheritance pattern has also been suggested. Patients generally present in the first decade of life. It affects males more than females. KT syndrome is a congenital circulatory disorder typically comprising of the triad cutaneous capillary angioma, bone and soft tissue hypertrophy and varicose veins. There are several theories about its pathogenesis. The management of this syndrome consists mainly of early diagnosis, prevention and treatment of complications. We report the case of a 30-year-old male patient with KT syndrome showing the classical triad.

Key words: Classical triad; magnetic resonance imaging; Klippel-Trenaunay syndrome; unusual age

Introduction

Klippel-Trenaunay syndrome is a congenital vascular disorder which is rare and affects one or both limbs. It has an incidence of about 2 to 5 in 1,00,000. French physicians Klippel and Trenaunay first stated this syndrome in the year 1900. They termed the syndrome "nevus vasculosus osteohypertrophicus"[1]. In 1907 Park Weber termed the same condition as hemangiectatic hypertrophy. The etiology is unknown. Though it is a sporadic condition, however paradominant inheritance pattern has also been suggested. Patients generally present in the first decade of life. It has predilection of affecting males more than females. Klippel Trenaunay syndrome is congenital circulatory disorder typically comprising of the triad cutaneous capillary angioma, bone and soft tissue hypertrophy and varicose veins. Pathogenesis has been described by several theories. The management of this syndrome mainly include careful diagnosis, prevention and treatment of complications.

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Case Report

A 30-year-old male patient was presented to our hospital with the complaints of dilated veins over the outer aspect of left lower limb since birth, which aggravated on standing and walking and was relieved on lying down or raising his limb. Physical examination showed hyperpigmented areas with raised margins (port wine stains) in the outer part of his left lower limb; extending from the mid-thigh to the mid-calf region [Figures 1 and 2]. His left lower leg showed multiple varicose veins. There was left lower limb hypertrophy with striking differences between various measurements in the lower limbs; such as: Length 82.5cm/87.5cm, mid-thigh girth 41 cm/48 cm, calf girth 31 cm/39 cm in the right and the left lower limb respectively. An incompetent saphenofemoral junction was demonstrated on the Trendelenburg test. Radiographs of the left leg showed a bony outgrowth arising from the lateral aspect of the upper end of the tibia and a deformed fibula. Cortical thickening was noted in the lower part of the shafts of the left tibia and fibula. Areas of cortical irregularity and marrow sclerosis were noted in the lower part of the shaft of the left fibula [Figure 3]. Ultrasonography showed hypertrophy of the soft tissue in the left leg; predominantly in the lateral aspect. Doppler sonography revealed incompetent left sapheno-femoral and popliteal junctions and showed multiple abnormally

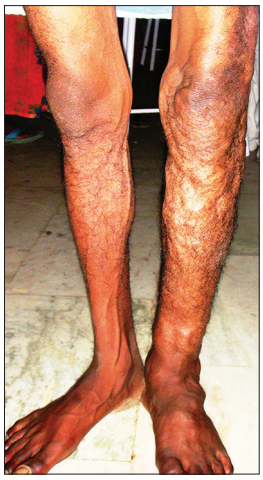

Figure 1: Hyperpigmentation (port wine stains) on the outer aspect of left lower limb extending from the mid-thigh to the mid-calf region

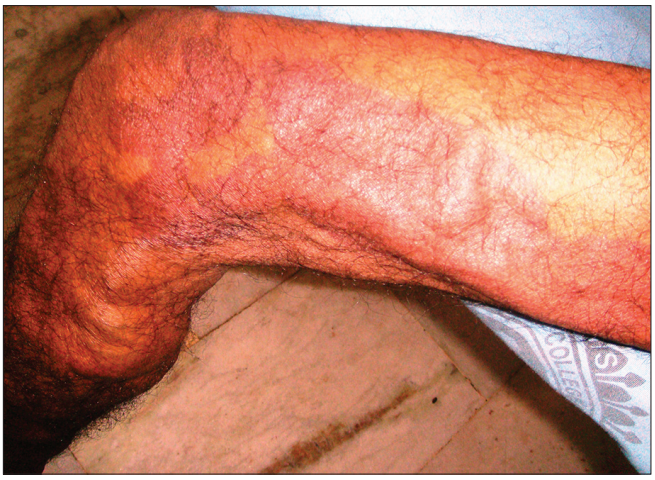

Figure 2: Hyperpigmentation (port wine stains) on the outer aspect of left lower limb extending from the mid-thigh to the mid-calf region

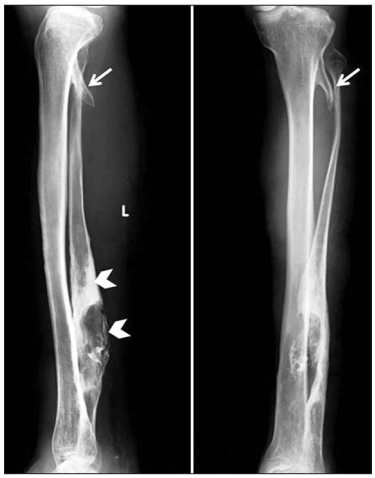

Figure 3: Bony outgrowth from the lateral aspect of the upper end of left tibia with deformed fibula. Cortical thickening involving the lower part of the shaft of both left tibia and fibula. Areas of cortical irregularity and marrow sclerosis in the lower part of the shaft of left fibula

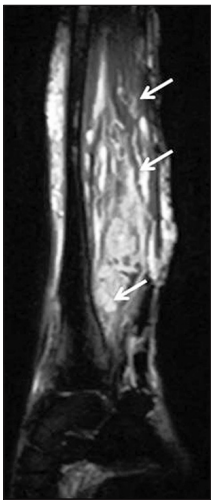

Figure 4: Marked hypertrophy of soft-tissues of the left leg with multiple abnormal vascular channels displaying altered signals, hyper intense on T2 suggestive of varicosities within the hypertrophied soft-tissue

dilated venous channels within the intramuscular and subcutaneous planes in the left leg predominantly involving the lateral aspect. Dilated perforator was noted in the popliteal region. Magnetic resonance imaging showed marked hypertrophy of soft-tissues of the left leg. Pressure effects over the tibia and fibula with focal areas of sclerosis and bone edema (hypo intense on T1 and hyper intense on T2) in the shaft of the left fibula, mainly in the lower half, was also noted. The hypertrophied soft-tissue showed heterogeneous signals hyper intense on T2WIs. Multiple abnormal vascular channels displaying altered signals, hyper intense on T2 and hypo intense on T1 (suggestive of varicosities) were seen within the hypertrophied soft tissue of all compartments involving the subcutaneous and muscular planes [Figures 4 and 5]. A diagnosis of Klippel-Trenaunay Weber syndrome was made on the basis of the triad including port wine stain, limb hypertrophy and lateral varicosity. Conservative treatment was given to the patient using elastic compression stockingsPatient was also advised to keep the limb elevated wherever possible and asked to come for yearly reviews.

Discussion

KT syndrome is also known as Angio-osteo-hypertrophy syndrome. It is a sporadic mesodermal abnormality that involves the capillaries, veins and lymphatics (combined vascular malformations) unusually distributed varicosities and enlargement of the limb. The age of presentation is usually at birth or during early infancy or childhood. The lower limbs are involved in 95 percent of cases, the upper limbs in 5 percent.

Capillary hemangiomata constitute the first presenting feature. They have deep violet color (in light-skinned people) with distinct linear borders and most commonly involve the lateral aspect of the affected limb. The capillary hemangiomata may be limited to the skin or extend deep to the subcutaneous

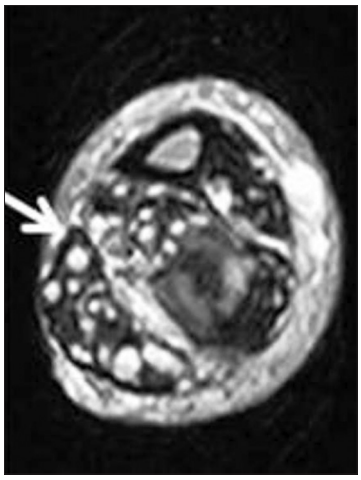

Figure 5: Marked hypertrophy of soft-tissues of the left leg with multiple abnormal vascular channels displaying altered signals, hyper intense on T2 suggestive of varicosities within the hypertrophied soft-tissue

tissue; and even involve the muscles and bones.[2] They are mostly unilateral and segmented and never cross the midline. Although the face and cervical region are the most commonly affected parts of the body, the proportion of the involved area increases with the child's growth and may include any part of the body. Lesions are usually light pink in infancy, but become progressively darker and finally become dark red as the child ages. Varicose veins, which result from defective valves, are often present at birth as large superficial veins extending from lower leg all the way to the buttocks. They may be extensive and spare the region of drainage of the saphenous vein. These affected areas may remain stable or gradually enlarge, leading to pain, lymphedema, thrombophlebitis and ulcers. Rarely, an arteriovenous fistula occurs in the affected limb; this feature helps to distinguish KT syndrome from Park Weber syndrome. Increased length and increased girth cause secondary limb hypertrophy. The digits may be affected initially and progress to eventually cause syndactyly, oligodactyly, polydactyly or macrodactyly. The cases in which soft-tissues (rather than bones) are predominantly affected may present with an increase in limb girth as the only feature. Gait disturbance is the initial sign of limb lengthening. Rarely, atrophy rather than hypertrophy occurs in the affected limb.[3] Hypospadias, hyperhidrosis, spina bifida, paresthesia, decalcification of affected bone, hypertrichosis, chronic venous insufficiency, dermatitis, venous ulceration and poor wound healing might be the other features of the disease.

Imaging plays an important role in the diagnosis and ongoing evaluation of this disease. Soft tissue thickening with bone elongation, extensive dilatation of superficial veins and segmental absence or hypoplasia of deep venous system may be found on a plain radiograph. MRI may reveal hypo intense signal on T1 and hyper intense signals on T2, which suggests clear delineation of venous and lymphatic malformation. The extensions of lesions and relationship to adjacent organ and structure can also be studied on MRI.[4] The evidence of superficial varicosities, enlarged perforating veins and absent or hypoplastic deep veins can be checked for on a MR venography. Its distinction from Park Weber syndrome can be done on angiography.

Surgery, sclerotherapy and compression therapy are used as treatment options. The effect of chronic venous insufficiency in the affected limb can be reduced by using graduated compression garments or intermittent pneumatic compression pumps may also be used. Thrombophlebitis and cellulitis can be managed with antibiotics, limb elevation, analgesia and corticosteroids. In cases with recurrent thrombophlebitis or before surgery, aspirin ± anticoagulants may be used as a prophylactic measure. Vein ligation, vein stripping, vein resection and amputation are the different types of surgical interventions, which are used.[5,6]

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