Klippel-Trenaunay Syndrome

N Forbes[1], M Walwyn[1], G Rao[2], D Ellis[2], MG Lee[3]

ABSTRACT

Klippel-Trenaunay syndrome (KTS) is a rare congenital, vascular disorder affecting one or more limbs. The syndrome is characterized by capillary malformations, soft tissue or bony hypertrophy and varicose veins or venous malformations. We present a case of this disorder in a twelve-year old boy who had an enlarged right lower limb with varicosities. Investigations revealed extensive superficial and deep venous varices, with dilatation of the right common iliac and external iliac veins. Klippel-Trenaunay syndrome should be suspected in a child presenting with capillary haemangioma and an enlarged limb.

Keywords: Congenital, limbs, vascular, veins

Síndrome de Klippel-Trenaunay

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RESUMEN

El síndrome de Klippel-Trenaunay (KTS) es un raro trastorno congénito vascular que afecta a una o más extremidades. El síndrome se caracteriza por malformaciones capilares, hipertrofia ósea o del tejido suave, y várices o malformaciones venosas. Presentamos un caso de este trastorno en un muchacho de doce años que tenía una extremidad inferior derecha agrandada con varicocidades. Las investigaciones revelaron varices superficiales y várices venosas profundas, con dilatación de las venas ilíacas comunes derecha y las venas ilíacas externas. El síndrome de Klippel-Trenaunay se debe sospechar en un niño que se presenta con hemangioma capilar y agrandamiento de un miembro.

Palabras claves: Congénito, extremidades, vascular, venas

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INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is a rare congenital malformation which is known by various names including angio-osteohypertrophy, nevousvaricosusosteohypertrophicus syndrome, hemangiectasiahypertrophicans and nevousverucosushypertrophicans (1). The combination of capillary malformations *ie* port wine stain (PWS), soft tissue and bony hypertrophy, varicose veins or venous malformations and lymphatic abnormalities characterize the Klippel-Trenaunay syndrome (2−4). This syndrome can be diagnosed if any two of the three features exist (2−3). The lower limb is involved in 95% of cases and the upper limb in 5% of cases (5). Less commonly, intra-abdominal and thoracic structures are affected (3, 5−8). Sporadic cases occur frequently but recently a gene has been associated with this syndrome (9).

CASE REPORT

A twelve-year old boy was admitted to the Cornwall Regional Hospital in August 2011 with a history of a painless enlarged right lower limb from early childhood. He also had a swelling in the right inguinal area that was consistent with

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varicosities. He did not have a port wine stain, did not walk with a limp and had no family history of the disorder.

On physical examination, the patient's vital signs were normal. Varicosities were noted in the inguinal region of the right lower limb and the latter was markedly enlarged compared to the left lower limb (Figs. 1, 2). Additionally, the

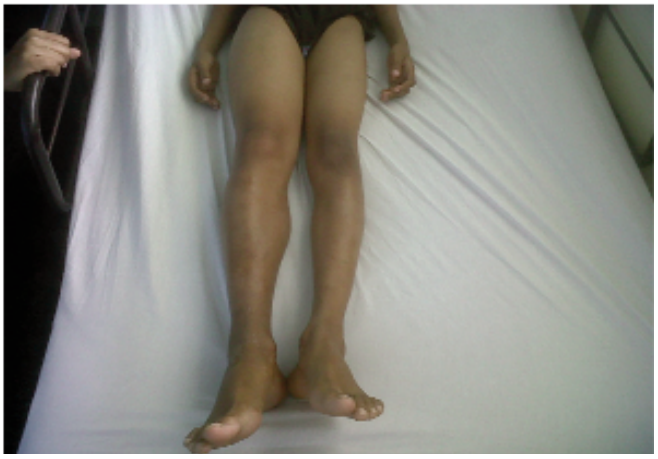

Fig. 1: Right lower limb hypertrophy.

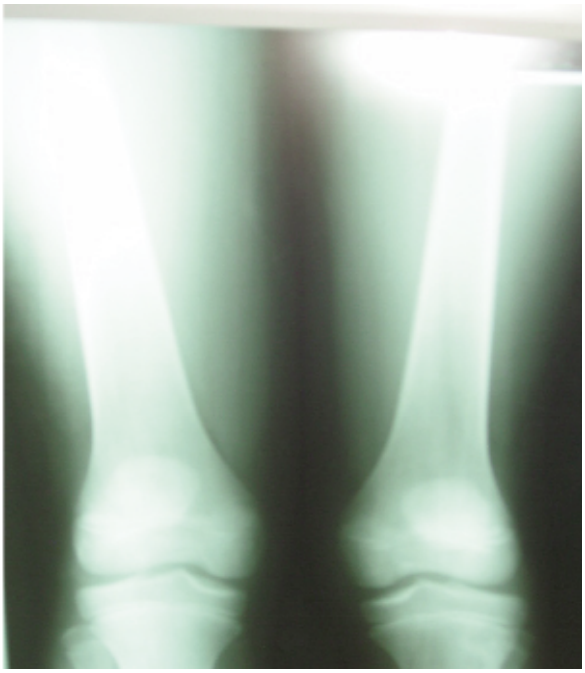

Fig. 2: Femurs and knees demonstrate soft tissue and bony hypertrophy on the right.

differences between both limb measurements were striking. The length of his right lower limb from the anterior superior iliac spine to the medial malleolus was 83 cm and the left lower limb was 80 cm. Thigh girth was 45 cm on the right and 36 cm on the left. The right and left leg circumferences were 33 cm and 25 cm, respectively. There was no evidence otherwise of hemihypertrophy and no developmental anomalies were noted.

Doppler ultrasound scan of the patient's right lower limb revealed prominent dilatation of the femoral and popliteal arteries on the right, proportionate enlargement of the associated veins and increased vascularity of the limb. There was also enlargement of the associated gastrocnemius and soleus muscles and right thigh muscles. A computed tomography (CT) of the right lower limb showed extensive superficial and deep venous varices with associated soft tissue hypertrophy and a right lower extremity that was significantly larger than the left. Additionally, the right common iliac and external iliac veins were also dilated. Varices were also demonstrated in the right hemipelvis and extended into the right femur. These findings were consistent with the Klippel-Trenaunay syndrome.

The patient was given conservative management with compression stockings and advised on yearly follow-up.

DISCUSSION

Two French physicians, Klippel and Trenaunay, were the first to describe the syndrome in the early 1900s in patients presenting with port wine stain, varicosities and osteohypertrophy of the affected limb (2). Parkes Weber also described the syndrome in 1907 as arteriovenous malformations associated with the above three findings (10). The syndrome was named Klippel-Trenaunay-Weber syndrome for a while but later the Weber was dropped to avoid confusion with the Parkes-Weber syndrome which also may present with an enlarged lower limb and has a poorer prognosis for limb salvage (11). Therefore, the Klippel-Trenaunay syndrome must be distinguished from Parkes-Weber syndrome, in which an enlarged extremity occurs that is related to an underlying arteriovenous malformation.

The syndrome usually occurs sporadically with rare cases having a familial predilection (11, 12). Both sexes are equally affected. The cause of the syndrome is unknown but several theories exist. One such theory is the presence of a mesodermal defect during embryogenesis causing the persistence of microscopic arteriovenous (AV) communications (13). Yet another theory postulates that intra-uterine damage to the sympathetic ganglia or inferiomedial lateral tract leads to microscopic AV communications (14). Two additional theories include loss of sympathetic nervous system regulation on end capillary flow and a neural crest phakomatoses with somatic anomalies (15). Through advances in modern genetics, a gene for KTS has been identified as angiogenic factor with G patch and FHA domains 1 [AGGF1] (9).

The clinical features are usually that of a haemangioma that appears first and at birth. The same may be present on any part of the body although the face and cervical region are most commonly affected. The midline is not crossed and the haemangioma does not involute but gets progressively larger with the growth of the child. Underlying structures may be

affected (16). Varicosities may present in infancy and usually progress and grow until adolescence. Limb hypertrophy is the last symptom to appear and the limb will grow until sometime in adolescence when growth is completed (17).

The syndrome is defined as the presence of capillary abnormalities, varicosities and bony and/or soft tissue hypertrophy. Two of the three features are sufficient for diagnosis. The present case presented with varicosities and bony and soft tissue hypertrophy involving the right lower limb demonstrated by both Doppler ultrasound and CT. He had no gait abnormalities, pain or other complications associated with the syndrome. Radiographic findings of the affected limbs of patients with KTS show soft tissue and bony overgrowth. Cortical thickening may also be present (6).

Klippel-Trenaunay syndrome should be suspected in a child presenting with capillary haemangioma and an enlarged limb. The differential diagnosis should include Maffucci syndrome, Beckwith-Weidemann syndrome, neurofibromatosis and macrodystophialipomatosa (12).

The syndrome is usually benign and management supportive unless complications occur. Complications may include bleeding, cellulitis, venous thrombosis, pulmonary embolism, hand and foot anomalies and lymphoedema. There is no curative treatment for KTS and management is symptomatic relief and treatment of complications such as bleeding, pain and length discrepancies. Pulsed laser dye therapy is used for port wine stains (18). Varicosities causing venous insufficiency are treated with compression stockings and surgical treatment may be used for superficial symptomatic varicosities (19). Complications such as cellulitis, deep vein thrombosis and superficial thrombophlebitis are managed medically (20). Surgical management may be necessary in some cases to correct uneven growth, debulk soft tissues and removal of excess veins or haemangioma (21). Possible procedures include osteotomy, epiphysiodesis and epiphyseal stapling (22). Follow-up monitoring should be done at least annually and intervention given if indicated (23).

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