Klippel-Trénaunay Syndrome

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Abstract

Klippel- Trénaunay syndrome (KTS) is an uncommon entity. This congenital malformation is characterized by the triad of soft tissue or bony hypertrophy, cutaneous vascular malformations, and atypical venous abnormalities. We report here a case of KTS and discuss the clinical features, investigations, and management of this enigmatic condition. ©

INTRODUCTION

Klippel- Trénaunay syndrome (KTS) is an uncommon yet distinct clinical entity characterized by a triad of soft tissue or bony hypertrophy, cutaneous vascular malformations, and atypical venous abnormalities. This congenital malformation should be correctly diagnosed and distinguished from the somewhat similar Parkes-Weber syndrome. Treatment is usually conservative.

CASE REPORT

A 13 year old boy presented with subacute onset of severe pain and swelling of the right calf. He had a long history of passing small amounts of fresh blood during defecation. He had no history of tuberculosis, bleeding diathesis, severe abdominal pain, worms in stool, trauma, or any other major medical or surgical illness.

On examination, he had severe pallor. His pulse rate was 110 per minute, and blood pressure was 104/70 mm Hg. He was afebrile, and did not have icterus, cyanosis, clubbing, lymphadenopathy, or edema.

He had a large patch of rough, thick, hyperpigmented skin on the anterior and lateral aspects of his right thigh (Fig. 1). This lesion had been present since birth and had not changed in size or appearance. His right calf was swollen and tender, and Homan’s sign was present. Thrombosed and varicose superficial veins could be palpated over his right calf. He had bony and soft tissue swelling of his right foot and syndactyly of 2nd and 3rd toes of right foot (Fig. 2). Rectal examination did not reveal any local pathology. No discrepancy in length was noted in the two lower extremities. Abdominal, respiratory, cardiovascular, and neurological examination was normal.

On investigation, his hemoglobin was 4.0 g/dL, with hypochromic, microcytic anemia. PT, aPTT, BT and CT were within normal limits. LFT and RFT were normal. ECG and X-ray chest were normal. X-ray of the feet revealed bony and soft tissue hypertrophy on the right side, along with syndactyly of 2nd and 3rd toes (Fig. 3).

Colour Doppler of the vasculature of the right lower limb showed complete thrombosis of the popliteal vein. Sigmoidoscopy showed multiple hemangiomas at 17, 15 and 10 centimeters from the anal verge. Mesenteric angiography detected a single hemangioma over the descending colon, distal to the splenic flexure. Barium enema was normal. MRI angiography of the right lower limb did not reveal any arterio-venous malformations. Chromosome analysis revealed a normal karyotype.

Thus this patient had a cutaneous malformation over his right thigh, right lower limb hypertrophy, right calf deep vein thrombosis (DVT), syndactyly of right 2nd and 3rd toes, and hemangiomas of colon. He was a classic case of the Klippel-Trénaunay syndrome (KTS).

He responded to treatment with blood transfusion, antibiotics, enoxaparin and hematinsics. Surgical consultation for management of colonic hemangioma was done; a conservative line of management was advised. He was asymptomatic at the time of discharge, and advised regular follow-up.

DISCUSSION

In 1900 two French physicians Maurice Klippel and Paul Trénaunay described a patient with asymmetrical hypertrophy of soft tissue and bone along with hemangioma of the skin.1 The Klippel-Trénaunay syndrome (KTS) is thus a triad of cutaneous capillary malformation, congenital venous abnormalities, and skeletal or soft tissue hypertrophy.

In 1907, Frederick Parkes-Weber described similar patients with both arterial and venous involvement. The Parkes-Weber syndrome (PWS) is a constellation of arterio-venous malformation (AVM), cutaneous capillary
malformation and skeletal or soft tissue hypertrophy.²

KTS occurs sporadically, and shows no particular racial, sexual or geographical predilection. It affects the skin, veins, lymphatic system, bone and soft tissue of an extremity.

252 patients (116 male and 136 female) with KTS were evaluated at the Mayo Clinic, USA, between January 1956 and January 1995.³ Port-wine stains were found in 98 percent, varicosities in 72 percent, and limb hypertrophy in 67 percent of these patients. 70 percent of the patients had involvement of the lower extremity, and four percent developed deep vein thrombosis. A minority of the patients displayed concomitant congenital anomalies like developmental dysplasia of the hip and syndactyly.

The cutaneous vascular malformation manifests as a flat reddish or purple capillary hemangioma. The skin may appear rough, thick, and hyperpigmented. Hemangiomas of the GI tract or kidney also may occur and cause bleeding per rectum. In the vast majority of children the skin lesion occurs on one leg, and is typically accompanied by lengthening and hypertrophy of the affected extremity.⁴

The cause of KTS remains obscure; many workers

believe that it occurs due to a sporadic mesenchymal abnormality in the development of the limb.⁵ Although the diagnosis is primarily clinical; confirmation requires laboratory and imaging studies.
Colour Doppler of arteries and veins and X-ray study of lower limb bones are obtained. Magnetic resonance angiography (MRA) has largely replaced the earlier methods of peripheral arteriography and venography.

Treatment is usually medical; only rarely is surgical or orthopedic intervention warranted.

Nonoperative management includes suitable footwear, graduated compression stockings and garments, and intermittent pneumatic compression pumps. Medical management includes antibiotics for cellulites, anticoagulants for deep vein thrombosis, and appropriate management of anemia. Oral contraceptives are contraindicated. Operative management is indicated for varicose veins and when the projected leg length discrepancy is more than 2 cm.

Acknowledgement
We wish to thank Dr. Archana Chowdhary, Professor and Head, Department of Medicine, and Dr. P H Shingare, Dean, Grant Medical College and Sir JJ Group of Hospitals, for permitting us to publish this case report.

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