

Klippel-Trenaunay syndrome: A case report

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Abstract

Klippel-Trenaunay syndrome is a rare disease characterized by the triad of port-wine stains, venous malformations or varicose veins and bone and/or tissue hypertrophy. Its etiology is not well defined and presents most often from birth. Treatment is usually conservative, and the interventions are limited to the treatment of complications. We report the case of a newborn with manifestations of the syndrome since birth.

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INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is a disease characterized by port-wine stains, varicose veins, and bone and/or soft tissue hypertrophy. It was first described in 1900 by researchers Maurice Klippel and Paul Trénaunay¹. It differs from Klippel-Trenaunay-Weber syndrome in which arteriovenous malformation is present².

The etiology is not well defined³. It is the result of a disorder in the embryonic development of the mesodermal tissues that affects angiogenesis at different stages. Another theory is that the obstruction and/or deep vein atresia causes chronic venous hypertension, resulting in port-wine stains, varicose veins, and limb hypertrophy⁴.

There is no documented sex predilection. In general, lesions are present at birth; in most cases, the disease manifests during early childhood¹.

It is a rare syndrome in Brazil and deserves attention since its early diagnosis is essential for proper treatment¹.

CASE REPORT

V.J.L.S, a 22-year old woman, G2P1A0, referred to the Clinical Hospital of the Federal University of Triângulo Mineiro (HC-UFTM), in the 29th week of gestation, with a provisional diagnosis of gastroschisis. An obstetrical ultrasound revealed a mass extending from the abdomen to the left lower limb. Magnetic resonance imaging showed an expansive formation, probably of vascular origin, extending from the left hemiabdomen to the left lower limb, at the leg level, with apparent infiltration of the myoadipose plane of the popliteal fossa and the pelvic cavity. The possibility of vascular malformation was considered.

The patient was monitored at the obstetric pathology clinic of the HC-UFTM, and underwent a cesarean section at term, giving birth to a male with a birth weight of 4,500 g. The newborn was referred to the neonatal ICU for monitoring and research.

On admission, the presence of port-wine stain was observed, extending from the flank and lower back to the left thigh and associated with a significant increase in limb volume and venous ectasia. The newborn underwent Computed Tomography angiography of the abdomen and pelvis, which showed increased volume of the left abdominal wall, retroperitoneum, right and left buttocks, left lower limb, and infrageniculate region compatible with vascular malformation.

The patient was monitored by the vascular surgery staff and diagnosed with KTS due to the presence of port-wine stains, venous ectasia, and limb hypertrophy. An expectant conduct was adopted for the syndrome.

DISCUSSION

KTS is characterized by the presence of capillary malformation associated with venous malformations or varicose veins, as well as bone and/or soft tissue hypertrophy. It occurs sporadically, although some familial cases have been reported. It usually involves the lower end of the body, but the trunk or face may also be affected⁵.

In most cases, both hemangiomas and varicose veins may be present at birth, as in the present case, but generally become more prominent until adolescence⁴. Lymphatic changes are observed in 70% of patients, manifesting as lymphedema, lymphorrhea, and susceptibility to cellulite.

Venous abnormalities include agenesis, hypoplasia, atresia, valvular incompetence, and occlusion of the deep venous system secondary to fibrosis⁶. Hypertrophy may be secondary to an increase in length (bone involvement) and/or increase in circumference (soft tissue involvement). It may also be present at birth, progressing during the first years of life⁴. In the present case, port-wine stains, venous ectasia, and soft-tissue hypertrophy were present at birth (Figures 1 and 2)⁴.



Figure 1. Port-wine stain, venous ectasia, and hypertrophy of the left lower limb can be observed.



Figure 2. Malformation involving only one limb is highlighted.

The diagnosis is essentially clinical⁶.

The main differential diagnoses are Klippel-Trenaunay-Weber syndrome, Maffucci syndrome, Proteus syndrome, and other capillary malformations not associated with any syndrome¹.

Associated complications include thrombosis, coagulopathy, pulmonary embolism, heart failure, hemothorax, and bleeding of abnormal vessels in the

gastrointestinal tract, kidney, or genitalia. Patients with abnormal lymphatic drainage are at greater risk of cellulite and infections⁴.

There is no curative treatment, and the therapeutic goals are to improve the patient's symptoms and correct the consequences of serious injuries and length discrepancy. In general, for capillary defects, the most common treatment method is "pulsed dye laser"; surgery is indicated when patients become excessively symptomatic¹.

Varicose veins are usually managed with conservative treatment, although surgery has been reported mostly in very symptomatic cases^{1,3}. Regarding bone hypertrophy, braces or surgery may be necessary to correct significant discrepancies in the length of the limbs¹.

Individuals with this syndrome should be closely monitored by a specialized team in order to avoid possible complications and receive proper treatment if they occur. The long-term treatment is conservative and an appropriate multidisciplinary approach is required, since the disease affects multiple organs⁷.

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