



KLIPPEL-TRENAUNAY SYNDROME : A CASE REPORT

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ABSTRACT

We present a rare case of a young patient with trenaunay klippel syndrome essentially affecting the disabling upper limb leading to radical treatment.

KEYWORDS: Klippel-Trenaunay syndrome - venous malformation – amputation.

INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is a rare congenital disorder first described by Maurice Klippel and Paul Trenaunay in 1900 as a triad consisting of cutaneous port-wine capillary malformations, varicose veins and hemihypertrophy of soft tissues and bone.^[1] The diagnosis is sometimes late, essentially clinical, but a radiological assessment of the vascular malformations is necessary before any treatment.

CASE REPORT

A male patient, 18 years of age, was developed, since the birth, a superficial venous circulation in the forearm of the left upper limb, the evolution was marked by the development and the propagation of this circulation taking the appearance of a vascular malformation, with the observation, from the age of 2 years, an overgrowth of the upper left limb relative to the right limb.

When the child was 12 years, he was hospitalized in the vascular surgery department, with a vascular malformation affecting the right upper extremity: the hand, forearm and half of the arm, enlarged with superficial venous dilatation and an increase in the temperature of the limb. Radiological investigations revealed bone overgrowth, and the presence of an enormous vascular malformation reaching the depth reaching the muscular plane with an arterial and venous component and the presence of numerous arteriovenous shunts. Since this hospitalization the patient has not received any follow-up until the age of 18 when the member became giant (Figure 1), with significant disability, and appearance of other localizations of the vascular malformation at the root of the patient, limb and left hemithorax gynecomastia and mass opposite the left

scapula (Figure 2 and 3), a CT angiogram was performed.



Figure 1: upper limb of our patient.



Figure 2: Left hemithorax gynecomastia.



Figure 3: Mass opposite the left scapula.

An amputation (Figure 4), under the tourniquet, of the upper third of the arm was carried out following the patient's request because of the major physical and psychological handicap.



Figure 4: amputation of the upper third of the arm.

The postoperative evolution was simple, with a good healing of the stump of the amputation and a good mood of the patient. A compression vest has been prescribed for the patient.

After five years, our patient has adapted live with a prosthesis without extension of venous malformation.

DISCUSSION

KTS is a rare congenital disorder first described by Maurice Klippel and Paul Trenaunay in the early 1900s and originally termed naevus vasculosus osteohypertrophicus.^[2]

The etiology and pathophysiology of this syndrome are

not known.

Several theories have been advanced to explain the etiopathogenesis of this complex syndrome. Baskerville et al. hypothesized a generalized mesodermal anomaly.^[3]

The existence of hypoplasias and venous aplasia has suggested that venous hyperpressure may be responsible for hypertrophy of the malformation.^[4]

Anomalies of embryogenesis have also been suggested. KTS may be related to abnormalities in vascular network development, during the embryonic phases of vasculogenesis and angiogenesis of members between the 3rd and 6th gestational weeks.^[5]

KTS should not be confused with Parkes Weber syndrome, which is defined by the triad of capillary malformations, limb disparity, and arteriovenous malformation.^[6]

KTS, although rare, is the most common syndromes angio-osteo-hypertrophic.

Although most cases of KTS are sporadic, there have been a number of familial cases reported. In most cases, a gain-of-function mutation in the catalytic subunit of the phosphatidylinositol 3-kinase gene is implicated in the pathogenesis, ultimately leading to proliferation and angiogenesis.^[7]

The treatment options for KTS are limited and mainly supportive.^[8]

Venous and lymphatic insufficiency leading to chronic edema is managed with compressive therapy and wound care, limb discrepancy is typically managed with shoe lifts and rarely managed surgically, and capillary malformations are treated with laser and/or ablative therapy if symptomatic.^[9]

To date, there have been no reconstructive surgical attempts for the venous insufficiency portion of the triad.^[10]

Our patient has started to expose long-standing debilitating problems secondary to the venous stasis disease of its upper left extremity. It has also been noted to have another location. The medical management of his left upper limb consisted of amputation without the possibility of another treatment, except compressive therapy, with stability of the disease to the present day (Figure 6).



Figure 6: healing of the stump of the amputation.

CONCLUSION

Although venous congestion is a known entity of KTS, the move to invalid consequences, as in our patient, traditional medical management fails to relieve symptoms and recourse to radical treatment will be necessary.

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