The Klippel-Trenaunay-Weber Syndrome. A case report and literature review.

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Summary

Association of bone and soft tissues hypertrophy, cutaneous haemangioma and superficial varicosity classically form the Klippel-Trenaunay-Weber syndrome. One case was diagnosed in a 12-year-old patient, admitted for a right lower limb congenital oedema. The goal of this study is to attract clinicians' attention upon this rare affliction with a difficult diagnosis and treatment. Clinically generalised oedema of the right limb was associated with voluminous superficial varicosity. A thrill was present at the internal side of the thigh, with a systolic murmur, indicating an arteriovenous fistula. Phlebography showed dilatation of deep venous network, compression of the right superficial femoral vein and doubling of left deep femoral vein. Muscular echography revealed the presence of a compressing mass of tissues at the thigh level, which motivated the surgical procedure. It was a muscle in abnormal position trapping the deep vein, which was freed after muscle resection. The patient died 4 days post operatively, from massive pulmonary embolism. Diagnosis of Klippel-Trenaunay-Weber syndrome must be considered when facing any congenital lower limb hypertrophy. Phlebography showing deep vascular lesions is an essential method of diagnosis. When surgical treatment is considered, the prevention of thromboembolic complications has to be instituted.

Key words: Klippel-Trenaunay-Weber syndrome, diagnosis, treatment.

Résumé.

Le syndrome de Klippel-Trenaunay-Weber associe classiquement une hypertrophie osseuse et des parties molles, un hémangiome cutané et des varices superficielles. Il a été diagnostiqué chez une patiente de douze ans, admise pour une grosse jambe droite congénitale. Le but de ce travail est d'attirer l'attention des cliniciens sur cette affection rare, de diagnostic complexe et de traitement difficile. Sur le plan clinique, on retrouvait un œdème généralisé du membre inférieur droit, avec de volumineuses varices superficielles. Un thrill était perçu à la face interne de la cuisse, ainsi qu'un souffle systolo-diastolique à l'auscultation, faisant suspecter la présence d'une fistule artério-veineuse. Au plan para clinique, la plhébographie montrait une dilatation du réseau veineux profond, un refoulement de la veine fémorale superficielle droite et, à gauche, une duplication de la veine fémorale profonde. L'échographie musculaire révélait la présence d'une masse musculaire compressive à la cuisse, laquelle avait motivé l'intervention chirurgicale. Il s'agissait d'un muscle en situation anormale, piégeant l'axe veineux profond, qui sera libéré après résection musculaire. La patiente est décédée au quatrième jour post-opératoire d'une embolie pulmonaire massive. Le diagnostic de syndrome de Klippel-Trenaunay-Weber doit être évoqué devant toute grosse jambe congéniale. La phlébographie, en objectivant les lésions vasculaires profondes, est un élément essentiel du diagnostic. En cas de traitement chirurgical, la prévention du risque thromboembolique doit être particulièrement rigoureuse.

Mots clés: Syndrome de Klippel-Trenaunay-Weber, diagnostic, traitement

Introduction

The Klippel-Trenauney-Weber Syndrome is a disease associating bone and soft tissues hypertrophy, cutaneous haemangioma and superficial varicosity. It is rare among vascular diseases. Lower limb oedema is often found in vascular pathology in Gabon. It is usually due to parasitic diseases in particular filariasis, whereas congenital lower limb hypertrophy is very rare. This study reports a case of congenital lower limb abnormality in a twelve-year-old girl operated upon at "la Fondation Jeanne Ebori", on whom the Klippel-Trenauney-Weber syndrome was diagnosed. The extremely rare occurrence of this pathology, the diagnostic problems that it brings and the difficulties in therapeutic treatment have motivated this report. Its purpose is to draw clinicians' attention on this affliction; the complexity of its diagnosis and therapeutic treatment.

Case Report

The 12-year-old girl was admitted into the Thoracic and Vascular Surgery Department of the "Fondation Jeanne Ebori" in Libreville on December 02/1997, for the treatment of a congenital right lower limb hypertrophy. During the investigations she complained of pain in the right lower limb on walking and standing. Examination of the right lower limb showed a generalised oedema of the leg and the foot with monstrous varicosity at the thigh-level (fig1).

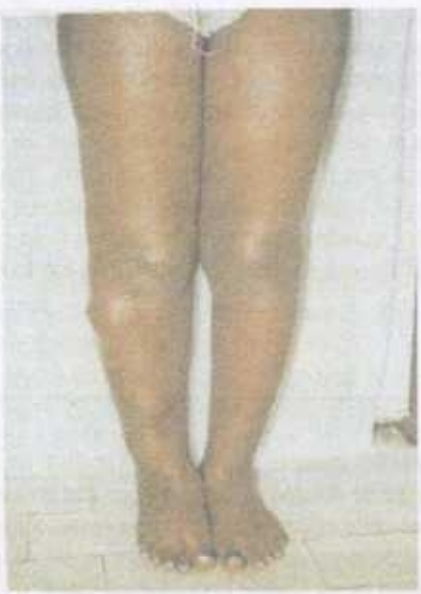

Fig 1. *Congenital fat right leg*
Visible subcutaneous varicosity of the right leg.
Note the difference of size between both the legs.

On palpation, there was no pitting oedema. There was a thrill noticeable at the medial side of the thigh. There was a round, firm painless swelling 3cm x 2 cm. The comparative measurements of the two lower limbs showed a gradient of 6 cm upper third, 13 cm lower third and 3.5 cm at the ankle (fig 2).

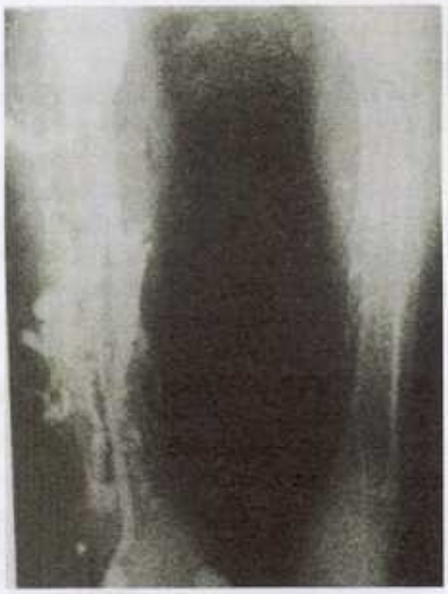

Fig 2. *Phlebography of lower limbs. Right deep venous system dilatation. Anterior and posterior tibial veins dilated*

Auscultation showed a systolic murmur at the internal side of the thigh. Upstream arterial compression to cause the disappearance of thrill in the presence of an arteriovenous fistula was negative. Other systemic examinations were normal. The sedimentation rate was 54 mm at the first hour and 100 mm at the second hour. The filariae test was negative. The lower limb phlebography showed an extensive venous network with the presence of posterior and anterior tibial varicosity. The huge popliteal vein measured 4.5 cm. The superficial femoral vein showed abnormality in its upper third (fig 3).

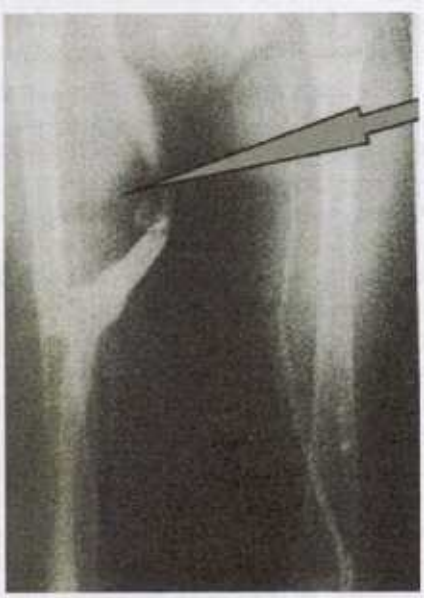

Fig 3. *Phlebography at femoral level Note the compression and deviation of the superficial femoral vein.*

The external vein collecting blood from superficial veins showed varicosity. The presence of an inverse flow phenomena in the curved segment and immediately downstream indicated the existence of an arterio-veinous fistula or of an extrinsic pressure by a mass. On the left limb, the deep superficial network was normal, apart from the presence of two deep femoral veins. The lower limb arterial blood supply was normal. The soft tissue scanning revealed the presence of an oblong tissular mass measuring 92 mm x 70 mm on the right at the thigh-level, with few calcifications. It was well delineated and was compressing the superficial femoral vein, causing statis and vascular distension. This was suspected to be a calcified haematoma or a myxoid tumour. The skeletal bone survey was normal. The electrocardiography showed a right ventricular hypertrophy.

The surgical indication was the compressive mass of tissues at the thigh level. The purpose of the surgery was to relieve the venous pressure, to reduce stasis, and improve the clinical development. At surgery, the adductor muscle was in abnormal position, located between the femoral artery and the femoral vein, compressing the femoral venous network. The femoral artery looked inflamed with thickened walls. The muscle was removed after cutting its upper insertion. On the fourth postoperative day, patient developed acute respiratory distress and died.

Discussion

The Klippel-Trenauney-Weber syndrome diagnosis was arrived at after elimination of other possible pathologies. The phlebitis was not considered and lymphangitis was eliminated due to the absence of corresponding inflammatory signs, temperature rise and regional lymphadenopathy. Investigation for micro filariasis was negative, but the scan was suggestive of a muscular tumour, or a calcified haematoma. The operation confirmed a femoral vein entrapment by a muscle in abnormal position, suggesting the popliteal entrapment syndrome, a well-known pathology in vascular surgery. The Klippel-Trenauney-Weber syndrome is characterised by a classical triad of skin vascular lesions, varicosity and bone and soft tissues hypertrophy. This triad is often associated with an arterio-venous fistula. It is now believed that one or two of the triad is enough for the diagnosis of this syndrome [1,2]. The absence of skin vascular lesions with haemangiomata can be explained by the patient's age, since these lesions disappear by the second and third years of age. The existence of an arterio-venous fistula is in other respect strongly positive. There was the presence of a typical thrill during the clinical examination and the observation of an inverse flow phenomenon during the phlebography. It is not surprising that the arteriography did not confirm the fistula. It is known that it is difficult to visualize arterio-venous fistula. It is the phlebography, which confirmed the diagnosis, showing features corresponding to those described in the literature for this pathology. Thus, the distension of the deep venous system and the connection of a huge external collateral at the thigh level, as in our case, suggested Klippel-Trenaunay vein [3]. Also the vascular compression by abnormal muscle fibrous strips, and the trapped femoral vein go along with the diagnosis [4]. Finally, the malformation such as that of the deep femoral vein doubling found in the phlebography, has been described in the Klippel-Trenauney-Weber syndrome [5]. The surgical treatment was the cutting of the muscle in abnormal position, and this relieved the venous compression. Surgery to reduce tissues did not seem to be indicated in this young patient, but a subcutaneous tissue liposuction was conceivable. Results of surgical treatment are not very satisfactory because of high rate of recurrence, poor healing and other complications [2]. The death of our patient seems to be due to a massive pulmonary embolism. Ischaemic necrosis of some organs is about a severe complication that has been reported in the literature [6]. These patients have a high risk of post-operative thrombosis, because the presence of many venous lakes outside the circulatory route contribute to this risk, by increasing the venous stasis. Adequate heparinisation, and the use of an umbrella filter or a vena cava clip will reduce the thromboembolic risk and prevent the mortality from pulmonary embolism.

Conclusion

The Klippel-Trenauney-Weber syndrome is a rare disease that must be considered in the presence of any congenital lower limb hypertrophy in a child, particularly if there is cutaneous haemangioma or superficial varicosity. The plhebography is a useful diagnostic tool. The treatment is complex and the results are usually not satisfactory. Thromboembolic phenomenon is very common.

Références

1. **KANTERMAN R.Y., WITT P.D., HSIEH P.S., PICUS D.** Klippel-Trenaunay syndrome: imaging findings and percutaneous intervention. Pictorial essay 1996, 167: 989-995.

2. **BASKERVILLE P.** Maladie thromboembolique et anomalies veineuses congénitales. Editions phlébologiques françaises 1966, 40 : 230-237.

3. **CHERRYK.,GLOVICZKI P., STANSON A.** Persistent sciatic vein: diagnosis and treatment of a rare condition. Journal of vascular surgery 1996, 23: 490-497.

4. **MEINE J.G., SCHWARTZ R.A., JANNIGER C.K.** Klippel-Trenaunay-Weber syndrome. From dermatology and pediatrics. New Jersey medical school 1997, 60: 127-131.

5. **AL-SALMAN M.M.S.** Klippel-Trenaunay syndrome: clinical features, complications, and management. Surgery today 1997, 27: 735-740.

6. **LANGERON P., TEILLAC HAMEL D.** Un Klippel-Trenaunay suivi de l'enfance à l'âge adulte : les problèmes de leur gestion. Phlébologie 1995, 48, 2 : 181-184.