Case Report

Case Report: Klippel-Trénaunay-Weber syndrome

Relato de caso: síndrome de Klippel-Trénaunay-Weber

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■ **ABSTRACT**

Introduction: The Klippel-Trénaunay-Weber syndrome (KTWS) is characterized by several signs, including capillary malformations and venous malformations with or without lymphatic malformations associated with limb overgrowth. In most cases, only one extremity is involved with arteriovenous malformation, and approximately 75% of the patients manifest symptoms before 10 years of age. **Case Report:** We report a case of a 7-month-old patient with KTWS followed-up at the Plastic Surgery Service of the Hospital de Clínicas, Federal University of Uberlândia; surgical treatment of the lesion was proposed for the patient. **Conclusion:** Since KTWS is a progressive disease with severe morbidity, the patient must be followed-up at a reference center by experienced staff with diverse therapeutic arsenal.

Keywords: Klippel-Trénaunay-Weber syndrome; Vascular malformations; Hemangioma.

■ **RESUMO**

Introdução: A síndrome de Klippel-Trénaunay-Weber (SKTW) é caracterizada pelo conjunto de sinais que consiste em malformações capilares, malformações venosas com ou sem malformações linfáticas associado ao supercrescimento de membros. Na maioria das vezes, envolve apenas uma extremidade com malformação arteriovenosa e cerca de 75% dos pacientes manifestam antes dos 10 anos de idade. **Relato de Caso:** Relatamos um caso de Klippel-Trénaunay-Weber em um paciente de 7 meses em acompanhamento na enfermaria da Cirurgia Plástica do Hospital de Clínicas da Universidade Federal de Uberlândia para o qual foi proposto tratamento cirúrgico da lesão. **Conclusão:** Como a SKTW é uma doença com morbidade progressiva e grave, o paciente deve ser acompanhado em um centro de referência com experiência e arsenal terapêutico diversificado para atuar da melhor forma possível no tratamento.

Descritores: Síndrome de Klippel-Trenaunay-Weber; Malformações vasculares; Hemangioma.

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INTRODUCTION

Klippel- Trénaunay -Weber syndrome (KTWS) is characterized by a set of signs that consist of capillary malformations, venous malformations with or without lymphatic malformations associated with limb overgrowth[1]. In most cases, it involves only one extremity with arteriovenous malformation and approximately 75% of patients manifest the disease before 10 years of age[2,3].

Klippel-Trenaunay syndrome was first described by Maurice Klippel and Paul Trenaunay. They reported two cases that had the triad (port wine stain, varices and bone and soft tissue hypertrophy) in common[4]. Some time later, Frederick Weber described some cases presenting a similarity to the triad, with the presence of arteriovenous fistula as an association[3,4]. SKTW and Parkes Weber's Syndrome may be considered together as Klippel-Trenaunay-Weber sd because they present different clinical signs of a single disease[5,6].

KTWS is a rare congenital mesodermal disorder, with about 1000 cases recorded worldwide[7]. KTWS is distributed equally among the various ethnic groups and affects more men, at a ratio of 1.5:1[7,8]. The etiology of this syndrome remains unknown, although there are some theories of its pathogenesis[9].

Although KTWS is a sporadic condition, studies have reported familial cases of KTWS that were not inherited through a Mendelian pattern, suggesting a multifactorial inheritance, with variable penetration and expression. Subsequent studies conducted by Happle suggest that the inheritance of a single defective gene acquired during embryogenesis could explain the development of this syndrome, as well as the occurrence of sporadic and familial cases, suggesting that an autosomal dominant inheritance is most likely[10,11].

Clinically, KTWS comprises flat hemangioma, venous changes such as malformations and varicose veins, and bone and soft tissue hypertrophy.[12,13]

The diagnosis is clinical and can be performed by the presence of the triad of abnormalities, or only two signs of the triad.

Usually, the patients with port wine stains, from birth, mainly in hypertrophied limbs, varying in depth[14]. Venous malformations affect the lower limbs in the vast majority of cases. Arterial or venous angiodysplasia may be present in any region of the body, from the skin to the visceral organs. Therefore, there is the possibility of phlebitis, bleeding, deep venous thrombosis, pulmonary embolism, hemoperitoneum, hemothorax and chronic venous insufficiency[15].

It is a rare syndrome, but deserves to be highlighted due to progressive and severe morbidity.

CASE REPORT

D.A.P., 7 months old, male, born and resident in Anápolis-GO, was forwarded to our service in the Emergency Room of the Hospital de Clínicas, Federal University of Uberlândia, MG, on 12/29/2014 with fever, vomiting, diarrhea and dehydration for 2 days. Together with this presentation, he presented a giant hemangioma in the left lower limb, varicose veins and hypertrophy of bone and soft tissues (KTWS) (Figure 1) with a history of recent laser treatment in another service of Goiânia-GO and with propranolol.

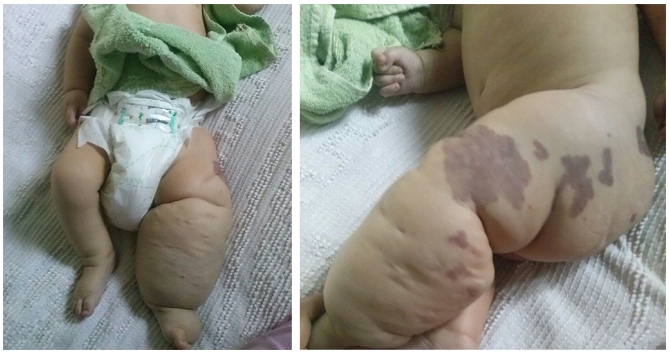

Figure 1. Giant hemangioma, varicose veins and bone and soft tissue hypertrophy in the lower left limb.

The patient progressed rapidly and severely with necrosis of the left lower limb (LLL) up to the ipsilateral gluteal region and with refractory septic shock (Figure 2). He received intensive ICU treatment with vasopressor drugs and broad spectrum antibiotic therapy for 28 days. Arterial and venous echo Doppler was performed during assessment of vascular surgery, confirming the diagnosis of Klippel-Trénaunay-Weber syndrome and identifying arteriovenous malformation and small fistulas of the left limb. However, extensive necrotic lesion in the LLL remained, requiring, primarily, surgical debridement of the LLL and subsequently left hip disarticulation with protective colostomy on 01/28/2015.

After 2 months of intensive clinical treatment, clinical and topical improvement of the lesion was observed in the postoperative region of the disarticulation(Figure 3A), and skin grafting was performed with electric dermatome and Mash Graft for occlusion of the bloody wound (Figure 3B), with the donor area of the right thigh. The patient presented good clinical recovery and epithelialization of the recipient and donor area, being discharged on the 15th postoperative day(Figure 3C).

The patient has been in outpatient follow up in this service, progressing satisfactorily with complete epithelialization of the donor and recipient areas.

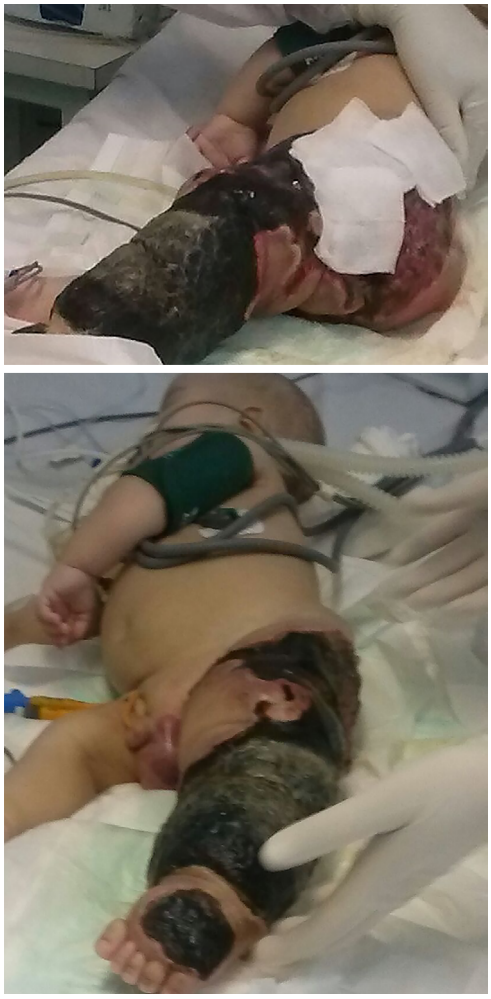

Figure 2. Presence of infection and necrotic tissue around the lower left limb up to the ipsilateral gluteal region.

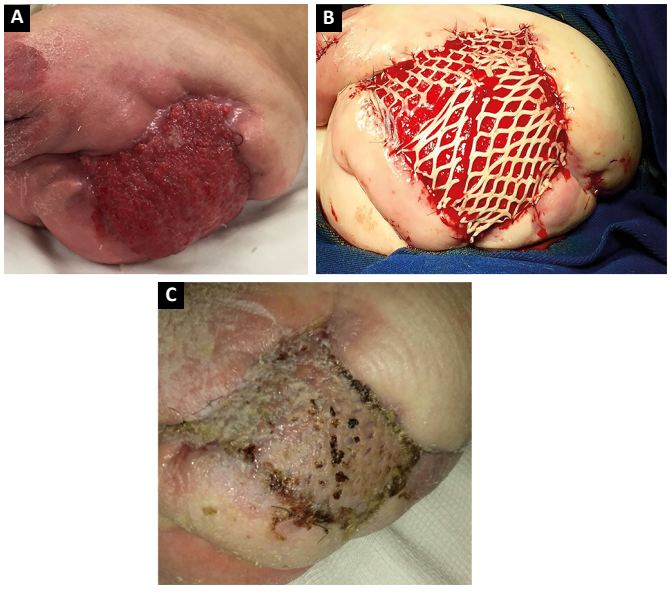

Figure 3. A: Bloody area after left hip disarticulation; **B:** Immediate postoperative aspect of mesh skin graft; **C:** 15 day postoperative aspect of receiving area.

The protective colostomy has been closed with the reconstruction of the intestinal transit.

DISCUSSION

This study was motivated by having received a patient referring prior treatment in another Plastic Surgery service with conservative treatment of KTWS with laser. Such conduct maintained the patient incapacitated, suffering with symptoms of chronic venous stasis and evolved with complications of local skin infection until receiving the final treatment.

There is no curative treatment, and the therapeutic goals are intended to improve the patient's symptoms and correcting the consequences of serious injuries and length discrepancy. However, all authors agree that conservative measures continue to guide the treatment of KTWS. This does not rule out the need for timely surgical interventions during the evolution of the natural history of the disease.

Adjuvant therapies can vary from laser therapy, with microfoam sclerotherapy, staged resections of ectatic veins and even larger excisions[1, 16-19] . The most commonly used indications for surgical treatment are: hemorrhage, local infections, thromboembolism and the occurrence of very refractory leg ulcers. Other indications are: local pain, functional limitations and aesthetics[17].

Interventional radiotherapy plays a major role in the propaedeutics of arteriovenous malformations (AVMs). Through it, one can evaluate the type of malformation and how the feeding vessels are structured. For the treatment of low flow AVM (KTWS), in some cases an injection of sclerosing agent may be applied to make the vessels smaller.

In other cases, this can be done using fluoroscopy. There are limited options for treatment of congenital venous dysplasia. In severe cases, the interventionist can use surgical removal, sclerotherapy or an endovascular ablation technique. If there are symptoms in the skin, such as Port wine stains, laser treatment may be indicated.

In cases such as the one described, considering the treatment performed, the area covered by grafting can be seen as a good reconstructive option, taking into account the simplicity of the procedure, less morbidity when compared to the use of flaps and the possibility of rehabilitation through the use of prostheses.

Despite being the highest level of lower limb amputation, the prosthesis is efficient, since the prosthesis for this level of amputation provides security and stability, with continuous gait, with or without locomotion aids, depending on other factors including the age of the patient.

KTWS should be suspected in all newborns with capillary malformations involving one extremity of the body from birth. The differential diagnosis for KTWS is

Proteus syndrome and Maffucci syndrome, among other non-syndromic capillary malformations[20].

Major advances will be made when it is possible to diagnose KTWS even more early and prevent the development of tissue hypertrophy, complex angiodysplasias and other phenotypic changes, perhaps by correcting or preventing the related genetic mutation[20].

CONCLUSION

KTWS is a rare disease, with progressive and severe morbidity. The patient with this syndrome must be accompanied in a reference center with experience and a diverse therapeutic arsenal to act in the best possible way in the treatment. Each patient has the parsimony and individualization in choosing the best treatment, as well as the ideal time to accomplish it.

Currently, the indication for surgical intervention is restricted to the complications arising from the initial presentation. In this case, the use of surgical treatment was crucial to allow an improvement in clinical condition and quality of life of the patient, showing that it can be a valid alternative.

COLLABORATIONS

VPB Writing the manuscript or critical review of its contents.

AOM Analysis and/or interpretation of data; writing the manuscript or critical review of its contents.

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