KLIPPEL-TRENAUNAY-WEBER SYNDROME

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Abstract: *Klippel-Trenaunay syndrome (KTS) is a congenital venous malformation that is characterized by the triad: varicose veins (generally affecting a single leg), hypertrophy of the affected limb and port-wine stain. Without treatment, the patients with KTS may develop chronic venous insufficiency with ulceration, massive bleeding, thrombophlebitis, pulmonary emboli, etc. In Parkes-Weber syndrome we can find arteriovenous malformations with fistulas. Some authors combine these 2 clinical entities in Klippel-Trenaunay-Weber syndrome. Establishing a correct diagnosis by assessing the existence of arteriovenous shunts and the types of this malformations as well as their severity, is important. We present 2 patients diagnosed in the Dermatology Department of the Clinical Hospital of Sibiu with Klippel-Trenaunay-Weber syndrome, complicated with repeated bleeding, undiagnosed for 12 years, respectively 20 years.*

Cuvinte cheie: *sindrom Klippel-Trenaunay, sindrom Parkes-Weber, malformaţii venoase*

Rezumat: *Sindromul Klippel-Trenaunay(KTS) este o malformaţie venoasă congenitală care ce se caracterizează clinic prin triada simptomatică: varice nesistematizate, frecvent unilaterale ale membrelor inferioare, hipertrofia globală a membrului afectat şi inconstant angiom cutanat. În lipsa unui tratament, KTS se poate complica (evoluţia varicelor spre insuficienţă venoasă cronică până la ulcer varicos cu posibilitatea apariţiei de hemoragii masive, tromboflebite, trombembolism pulmonar, etc). Sindromul Parkes-Weber asociază tabloului clinic anterior fistule arterio-venoase, malformaţii vasculare şi angiom gigant, de tip cavernos. Unii autori reunesc aceste 2 entităţi clinice în sindromul Klippel-Trenaunay-Weber. Stabilirea unui diagnostic corect, cu aprecierea imagistică a existenţei şunturilor arterio-venoase şi a tipurilor de malformaţii venoase şi arteriale ca şi a severităţii acestora este foarte importantă. Lucrarea de faţă prezintă cazurile a 2 pacienti în vârstă de 20 de ani, respectiv 40 de ani diagnosticati în Clinica de Dermatologie a Spitalului Judeţean Sibiu cu sindrom KTW complicat cu hemoragii repetate, nediagnosticat timp de 12 ani, respectiv de 20 de ani.*

CASE PRESENTATION

Case 1

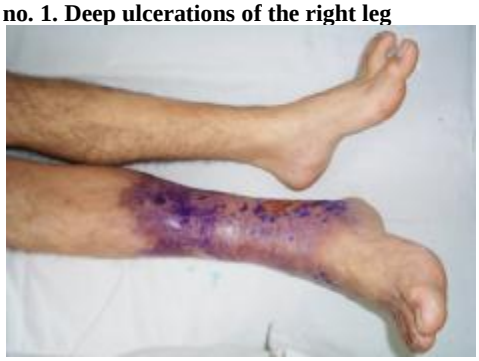
We report a 20-year-old male hospitalized in the Clinic of Dermatology of Sibiu for large, deep and painful overinfected ulcers (figure 1), important varicose veins (figure 2) and hypertrophy on the right leg with static and walking difficulties (figure 3). The symptoms became evident around the age of 8 years old by the appearance of varicose veins on the right leg. In progress appeared pigmented and purple dermatitis with ulceration on the right leg. Over the past 12 years the patient presented repeated episodes of bleeding from the ulcerated varices. During growth and development of joint and bone system it appeared a right leg hypertrophy.

At the moment of admission the difference between the circumference of the right and left thigh was 9 cm. At the leg level the difference was 6 cm (figure 4). Also, there was a difference between the lengths of the legs of 7 cm.

Laboratory investigations were within normal limits and the bacteriological exam from the ulcer did not reveal germs. Hypersfigmia in the lower limbs was evidenced at oscillometric examination.

The radiography of the pelvis shows calcification of soft tissues in the right groin area and spina bifida occulta of S1 (figure 5).

Figure no. 1. Deep ulcerations of the right leg



The Doppler ultrasonography of the arterio-venous system of the right lower limb revealed an important dilatation of the femoral vein with turbulent, arterial flow inside it, uninfluenced by the respiratory phases. Also, we found a significant dilatation of the superficial venous system with extensive varicose veins in the thigh and a small communication between the femoral artery and the femoral vein. The femoral artery presents a normal flow.

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Figure no. 2. Varicose veins of the right leg

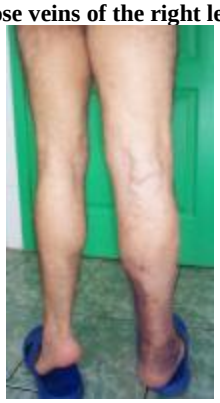


Figure no. 3. Important hyperthrophy of the right leg

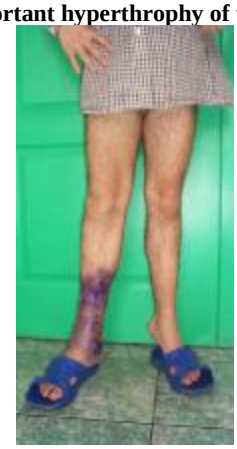


Figure no. 4. Muscle hypertrophy of the right leg

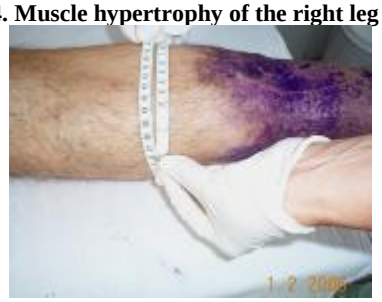
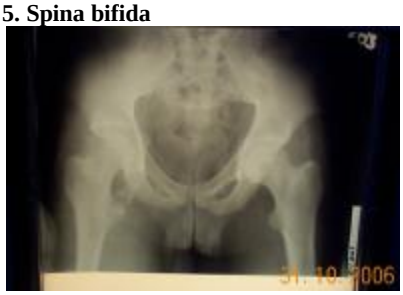


Figure no. 5. Spina bifida



The ultrasound diagnosis was arterio-venous fistula, probably in the context of vascular malformation and secondary varicose veins in this area.

The oscillometric hypersfigmia was suggestive for arterio-venous anastomoses (1). This aspect was concordant with the result of the Doppler ultrasonography (arterio-venous fistula).

Due to a history of repeated episodes of bleeding from ulcers and the bleeding risk during surgery we recommended a conservative treatment (non-elastic bandages associated with flebetonic drugs. We chose the non-elastic bandages compression because this form of compression provides a higher pressure during muscle activity and a lower pressure during resting with the possibility of keeping bandages for several days. Through this therapy we obtained the epithelization of the ulcer after 4 months.

By bringing together clinical data (varicous veins, hypertrophy of the right lower limb, the absence of cutaneous angioma, static and walk disorders) and workup data (Doppler ultrasonography and oscillometric exam) we interpreted the case as Klippel –Trenaunay-Weber syndrome.

PARTICULARITY OF THE CASE

- Young patient, undiagnosed for 12 years with severe vascular congenital malformation complicated with episodes of repeated bleeding from ulcers.
- ***Simple Doppler ultrasonography allowed the diagnosis.***
- At the moment of the diagnosis there were complications (chronic venous insufficiency with ulcers, repeated episodes of bleeding from the ulcerated varicose veins, static and walk dysfunctions of the right lower limb).
- The association of Klippel-Trenaunay syndrome with spina bifida (encountered also in our patient) is recognized in the literature.
- The history of episodes of repeated bleeding from the ulcerated varicose veins required the association of medical treatment with non-elastic compression bandages and general recommendations (to avoid prolonged standing, the local trauma and to make a correct ulcer dressing).

Case 2.

We report a male patient with KTWS, diagnosed 20 years ago. The late diagnosis allowed the evolution in a severe form of the disease with significant hyperthropy in lenght and thickness of the left leg with static and walking difficulties (fig 6).

Figure no. 6. The clinical aspect after 20 years from the diagnosis

DISCUSSIONS

The exact etiology of the KTWS is not fully known, but some authors suggest that the disease is due to a mesodermic anomaly during the fetal development, that causes the arterio-venous shunts to persist in the early lower limbs (2). Other authors consider that KTWS is caused by a genetical mutation (in the RASA1 gene which encodes the triggering protein GTP – 120-Ras).

In 1988 Young establishes a difference between these two entities (K-T syndrome and P-W syndrome) (6). We present these differences in the table below.

	KTS	PWS
Skin color in above of vascular malformations	Blue-violaceous	Pink, diffuse
Arterio-venous fistula	unimportant	important
Venous anomalies	frequent	-
Site on the upper limbs	5%	23%
Site on the lower limbs	95%	77%
Hypertrophy of the affected limb	Often disproportional, with the involvement of the soft tissues and the bone structures; often macrodactyly at the foot	Difference in the length of the limbs
Prognosis	Favorable; sometimes complications like pulmonary thromboembolism may appear.	Not favorable; bradycardia and cardiac insufficiency may develop.

Today, these two clinical entities are gathered in the KTWS.

The triad of symptoms in KTWS contains:

- ***cutaneous angioma*** (rarely subcutaneous, muscular or in the internal organs)
- ***varicosities and vascular malformations with arterio-venous fistulas***
- ***muscle and bone hypertrophy of the involved limb*** (more evidenced in patients with arterio-venous malformations).

The cutaneous hemangiomas in KTWS are in most cases plane, but sometimes one can meet also cavernous angiomas and lymphangiomas.

The varicosities may be evident from birth, first at the lower part of the leg, with progressive advancement, or later during the child-period.

The hypertrophy of the involved limb may affect its length (through bone involvement) or/and its width (through soft tissue involvement).

Due to the vascular malformations, in the skin one can develop chronic venous insufficiency with ulcerations, skin atrophy, cellulites. Other possible complications of KTWS are: thrombophlebitis, hemorrhages, pulmonary thromboemolism, scoliosis, paralysis, angiosarcomas. In the medical literature one can find numerous diseases, which are more frequent in association with KTWS: spina bifida, hypospadias, polydactyly, hyperhydrosis, hypertricosis, loss of bone tissue, orofacial malformations (3).

Necessary diagnostical investigations are osscilometry, Doppler ultrasonography, phlebography, arteriography, lymphography, MRI, CT-scan, bone-radiography (4).

The vascular malformations may also be present in the digestive- and the genito-urinary tract, with secondary hemorrhages at these sites (2).

The therapy of KTWS is challenging. The compressive treatment has appropriate effects in the management of chronic venous insufficiency, as well as protective effect against local trauma. The surgical treatment of the arterio-venous insufficiency is still a controversial subject (in the absence of complete investigations, surgery can increase the number of complications). Besides the compressive and phlebotonic treatment, endovenous therapy of the safena magna is an efficient and less invasive therapeutical alternative (5).

CONCLUSIONS

- Patients with KTWS need periodically clinical follow-ups, in order to prevent complications.
- In our patient, the lack of diagnosis over a period of 12 years, allowed the disease to evolve into chronic venous insufficiency with multiple ulcers of the right lower leg, repeated episodes of hemorrhage from the ulcers and dysfunctional walk and static of the right lower leg.
- Because of the past episodes of hemorrhages from the ulcers, we chose to apply the non-elastic compressive therapy, in association with phlebotonic drugs, which allowed the healing of the ulcerations and the improvement of the chronic venous insufficiency.

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