Parkes-Weber Syndrome with Pulmonary Hypertension Due to Multiple Arteriovenous Fistulas

Parkes-Weber syndrome (PWS) is an arteriovenous vascular malformation. It occurs at birth and affects mainly the lower limbs (77%). Detection of arteriovenous fistula associated with the Klippel-Trénaunay syndrome (KTS) triad confirms the diagnosis of this syndrome. (1, 2)

The KTS is characterized by the typical triad of cutaneous vascular port wine stains, soft tissue and/or bone hypertrophy and varicose veins (Figure 1). The difference between PWS and this syndrome is that PWS presents high-flow vascular lesions and arteriovenous fistula, no anomalous lateral veins, lymphatic malformations are rare, and the musculoskeletal involvement and dissymmetry of the affected extremity is lower than in KTS. (3)

The major complication in PWS is the increased cardiac output that can lead to heart failure, pulmonary hypertension, and cutaneous ischemia, caused by high-flow arteriovenous fistulas. (3)

Although its etiology is unknown, PWS has been associated with mutation of the E133K gene in the angiogenic factor VG5Q, which controls the growth of blood vessels for angiogenesis and vasculogenesis during embryonic development, and is transmitted by autosomal dominant inheritance with variable expressivity. PWS is much less common than KTS. (4)

High cardiac output with heart failure and pulmonary hypertension is associated with different conditions, including traumatic arteriovenous fistulas and therapeutic and congenital ductus arteriosus. The primary pathophysiological event is reduced peripheral vascular resistance due to peripheral vasodilation or arteriovenous fistula, and both scenarios can lead to a reduction of systemic blood pressure, neurohormonal activation, ventricular remodeling, and heart failure.

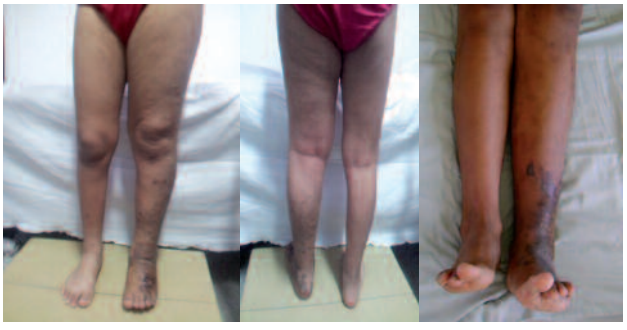

Fig. 1. Echocardiographic image following self-expandable valve implantation.

In these cases, conventional therapy for heart failure, such as angiotensin-converting enzyme inhibitors, beta blockers, and angiotensin receptor blockers, reduces peripheral vascular resistances and may result in the patient's clinical deterioration, although the literature reports this treatment in some cases.

Treatment consists of elastic compression, ligation of arteriovenous fistulas, orthopedic therapy, and eventually percutaneous embolization, in many cases resulting in limb amputation.

We present the case of an 11-year-old male Argentine patient, with no family history of this condition. The patient presented with an 8-year history of the disease, with difficulty in walking during childhood associated with multiple falls, increased volume, hyperpigmentation and indurations in the left lower limb.

He referred FC II dyspnea (New York Heart Association [NYHA] classification) associated with nocturnal dry cough. He also presented with elevated lesions and pruritic honey-colored scabs, moderate pain and heaviness of the left lower limb together with spontaneous hemorrhages described as a continuous stream of blood shooting up to 50 cm, with a frequency of five episodes in the last month.

The patient was oriented and afebrile; BP in right arm: 100/60 mm Hg; BP in left arm: 100/50 mm Hg; HR: 94/min; T: 36.6 °C.

Cardiovascular system: Jugular venous distension at 0 plane level, normal S1; regular S2: (P2 > AO2), no S3, non-irradiated systolic murmur in pulmonary focus 2/6, apex beat in 5th LIS.

Limbs: Left lower limb: increased volume, 11 cm in the thigh and 6.6 cm in the leg, 5 cm in the ankle, and 3 cm high with respect to the contralateral limb; increased temperature, multiple isolated brownish macular lesions of net, irregular borders, indurated hyperpigmented lesions ('port wine' stains), and visible venous pathways; macules, peripheral scaling and multiple blood stretch marks were observed in the dorsum of the ipsilateral foot.

Murmur with thrill in the dorsum of the left foot; pulses, altered ambulation.

Chest X-ray: CTI > 0.50.

Electrocardiogram: RS HR: 84/min, axis at 40°, left atrial and ventricular overload.

Color Doppler echocardiography: LVDD: 56 mm, LVSD: 32 mm, IVS: 10 mm, PW: 7 mm, Ao root: 26 mm, LA: 38 mm, RVDD: 18 mm, EF 70%, ShF: 42%. Pulmonary annulus: 26 mm, inferior vena cava: 15 mm, collapse <50%, TAPSE: 23. Left chamber dilation; preserved biventricular function. Moderate pulmonary hypertension, PASP: 50 mm Hg.

Arterial and venous lower limb Doppler ultrasound: Calibers: FV: 10.9 mm, PV: 10.8 mm, PV: 4 mm, CFA: 8.8 mm, PA: 7.6 mm, TPI: 4.6 mm, pedial artery: 3.36 mm. Multiple high-flow arteriovenous fistulas in CFA, proximal and distal popliteal artery, and at the tibial-peroneal trunk. Impaired venous flow, increased density and velocity were found on pulsed Doppler. Right lower limb: Without vascular disorders.

Multislice computed tomography angiography of the lower limbs: Hypertrophy of the left lower limb is observed. During scan acquisition of the arterial phase, opacification of the superficial and deep venous drainage system is seen, suggestive of microfistulas or fistulas (Figure 2).

Impaired bone structure at the calcaneal and mid third of the calf bones is found, as part of the vascular disorder.

Varicose dilatation of the superficial venous drainage system is observed. No signs of superficial and deep vein thrombosis are detected.

Changes are appreciated in the density of subcutaneous tissue of the leg, associated with edema.

Right lower limb: No vascular disorders are detected. Bone structures and soft tissues are normal.

The case reported matches the clinical data and imaging findings characteristic of this syndrome, together with the musculoskeletal hypertrophy of the left lower limb and increased circumferential and longitudinal diameter, cutaneous vascular disorders and high-flow arteriovenous fistulas in popliteal, femoral, and tibial vessels demonstrated with venous Doppler ultrasound and multislice CT angiography (Figure 3). (5)

Over the course of time, high-output arteriovenous fistulas cause dilation of the heart chambers, pulmonary hypertension, and heart failure, (6) which is the most severe complication. The effective fistula treatment is essential to prevent this hemodynamic abnormality.

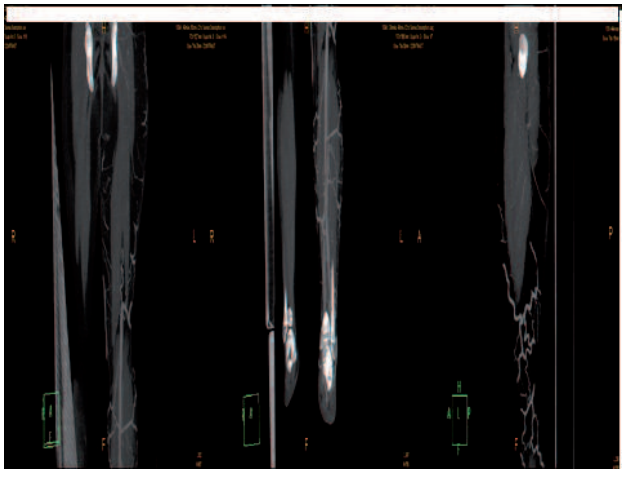

Fig. 2. Echocardiographic image following self-expandable valve implantation.

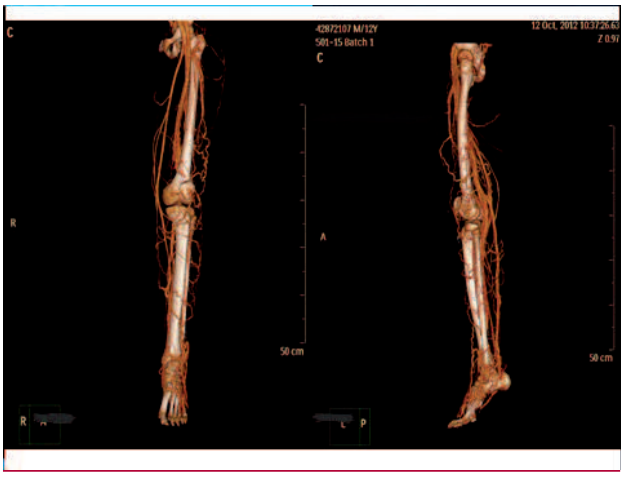

Fig. 3. Echocardiographic image following self-expandable valve implantation.

Conflicts of interest

None declared.

(See authors' conflicts of interest forms on the website/Supplementary material).

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