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Parkes-Weber Syndrome with Spinal Arteriovenous Fistula in Childhood

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A 9-month-old male infant with an enlarged left lower limb associated with port-wine stain over the left leg and buttock was diagnosed as Klippel-Trenaunay syndrome (KTS). He presented with progressive weakness in the lower limbs, evolving to paraplegia. Imaging studies showed arteriovenous malformation in the scrotum and high-flow fistulas in the left lower limb and spine, with ectatic veins and venous aneurysm compressing the spinal cord as shown in Figure 1. These imaging findings were consistent with Parkes-Weber syndrome (PWS), distinct from KTS [1]. KTS features consist of port-wine stain (capillary malformations) and varicose veins, associated with soft tissue and bone hypertrophy affecting lower limb in 95% of the cases; in addition, KTS is usually associated with low-flow lesions secondary to mutation of the AGGF1 gene, with relatively good prognosis [1–3]. In contrast, PWS results from mutations in RASA1 gene, which produces high-flow arteriovenous fistulas with consequent worse prognosis than KTS [3, 4]. Whereas only KTS is associated with limb hypertrophy, similar features of cutaneous stain may be found in several vascular anomalies, including KTS, Cobb syndrome, Sturge-Weber syndrome, and capillary malformation-arteriovenous malformation [2].

PWS is frequently misdiagnosed as KTS, due to the overlapping features of cutaneous stain and limb hypertrophy, or even incorrectly called Klippel-Trenaunay-Weber syndrome [2]. However, the distinction between these syndromes is relevant because their prognosis and treatment may differ significantly [1]. Besides clinical features, radiological noninvasive diagnosis with MRI projection angiography, as well as genetic testing, is a useful tool to evaluate patients with vascular malformations and limb overgrowth [1, 4]. High-flow fistulas and clear venous hypertension should be treated to avoid hemorrhage or mass effect as seen in our patient [2]. Therapeutic options for PWS patients include endovascular embolization and surgical resection, and multidisciplinary teams improve patient care. It allows individualized treatment based on risk-benefit analysis according to the vasculature characteristics and lesion behavior [2]. In our case, endovascular treatment was performed through the artery of Adamkiewicz, and the fistulous point was occluded with cyanoacrylate glue (Glubran, GEM Srl,

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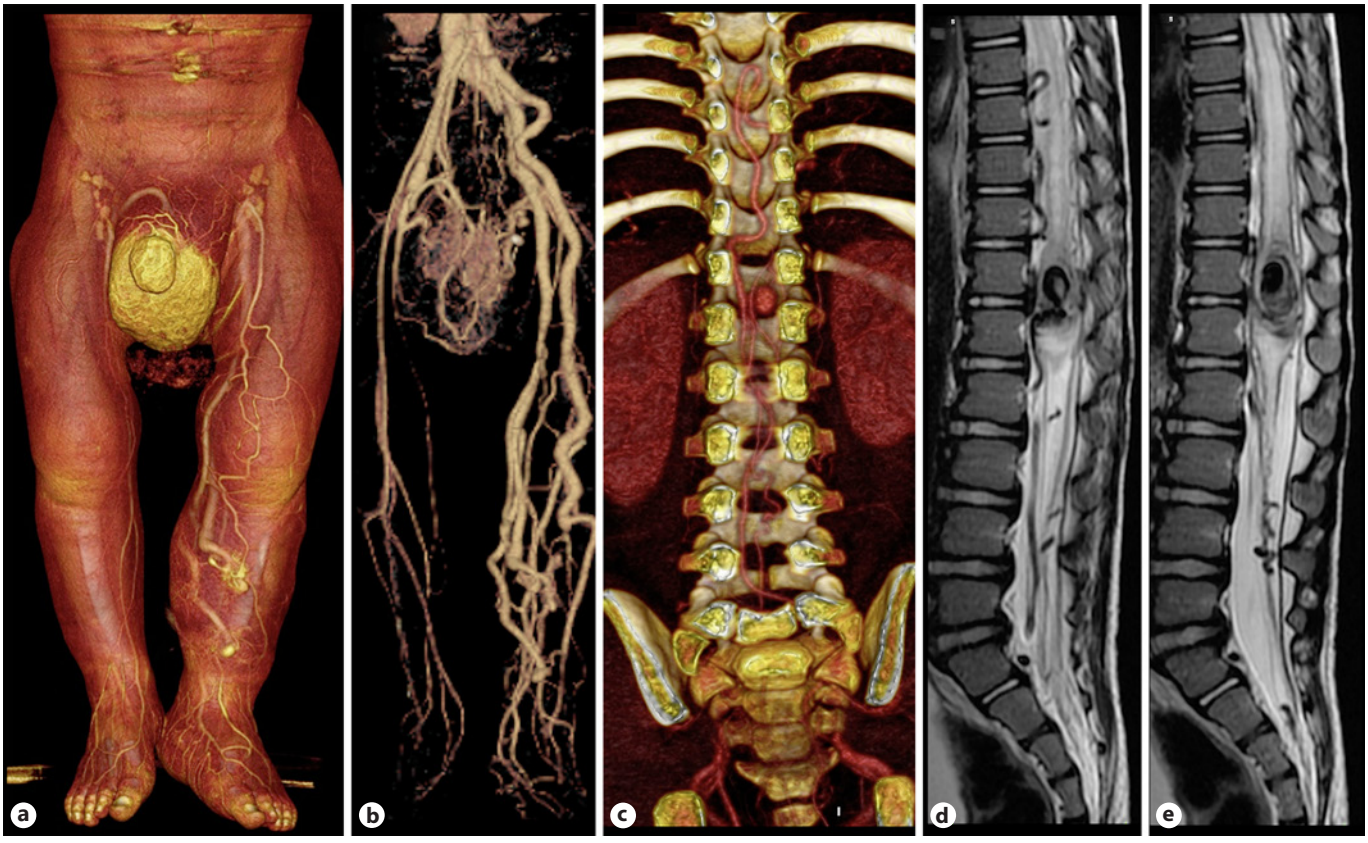

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Fig. 1. A 3D CT scan (**a**) and CT angiography (**b**) in an infant presenting with paraplegia show left inferior limb hypertrophy associated with arteriovenous malformation of the scrotum; the spinal CT angiography (**c**) and MRI (**d**, **e**) show an arteriovenous fistula, with enlarged draining veins and a venous aneurysm compressing the spinal cord.

Viareggio, Italy) diluted with Lipiodol (Laboratoire Guerbet, Aulnay-Sous-Bois, France) (Fig. 2a–c). The patient had progressive recovery of lower limb strength, being able to walk. Follow-up angiography and MRI showed durable fistula occlusion (Fig. 2d–f); however, the artery remained ectatic due to additional arteriovenous malformation. Three years later, the scrotum lesion required treatment due do ulceration and bleeding, confirming the aggressive course of PWS.

Statement of Ethics

This study was conducted ethically in accordance with the World Medical Association Declaration of Helsinki. Written informed consent for publication of data and images was provided and signed by the patient's guardian.

Conflict of Interest Statement

The authors declare no conflicts of interest.

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Author Contributions

Z.D.J. and A.K.M.: conception of the work and acquisition of the data; G.L.K., L.A.M.G., and A.A.C.D.: analysis and interpretation of the data; Z.D.J. and A.K.M.: drafting of the work; all authors: critical revision of the work for intellectual content and final approval of the version to be published. All authors are responsible for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

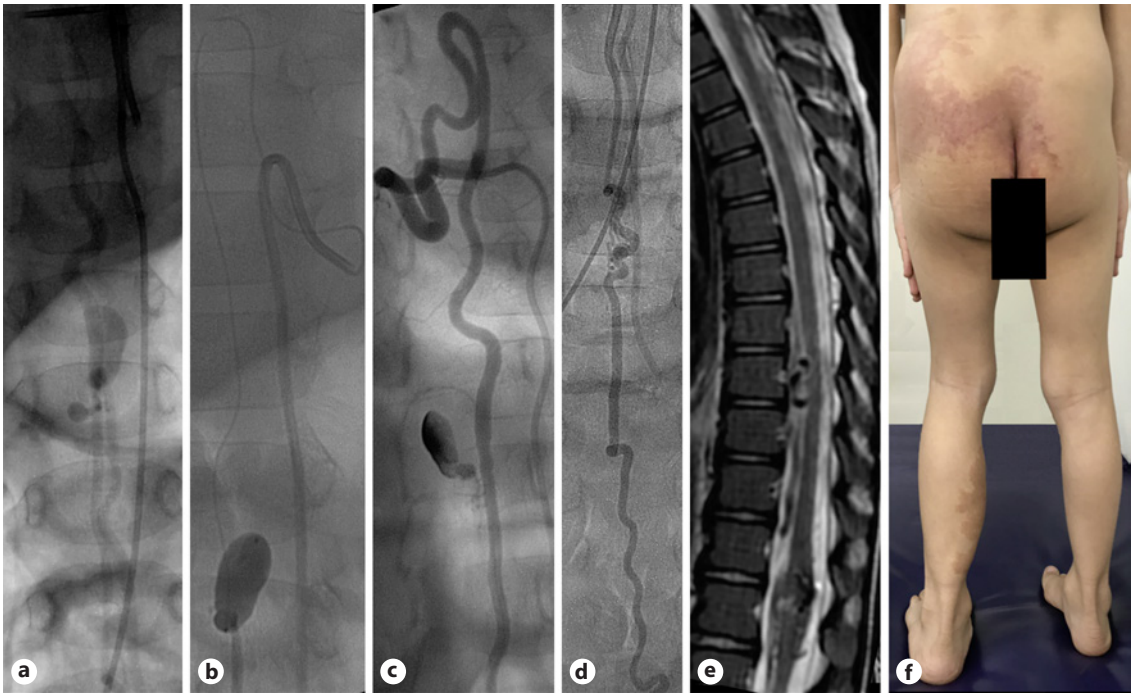

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Fig. 2. The angiography of Adamkiewicz's artery allows selective catheterization of the arteriovenous fistula (**a**, **b**); the fistulous point is occluded with glue, resulting in contrast stagnation inside the venous aneurysm, maintaining the distal arterial filling (**c**); the patient recovers the lower limb strength, and follow-up angiography shows durable fistula occlusion (**d**), while the MRI shows reduction of the spinal cord lesion (**e**); in 4 years, he is able to walk, and the picture shows the port-wine stains over the buttock and the hypertrophic left leg (**f**).

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