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Case report

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Diagnosis of Parkes Weber syndrome affecting a newborn baby´s upper left limb in a low resource setting: a case report

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Abstract

Vascular malformations (VMs) is a term used to describe vessels developmental abnormalities. These malformations involve arteries, veins, capillaries or lymphatics vessels. It can result to high-flow arteriovenous malformations (AVMs), associated with shunting of large amounts of arterial blood into the venous system; these malformations can lead to venous engorgement, and lately to high-output cardiac failure. Parkes Weber syndrome (PWS) is a form of VMs in which we have significant arteriovenous shunts. Patient with PWS have dilated and frequently visible pulsate varicose veins with other visible signs of arteriovenous shunt. There is frequent bones involvement. PWS is still frequently misdiagnosed as Klippel-Trenaunay syndrome, which is a triad of malformations involving the capillary, venous, and lymphatic vessels, without an arteriovenous fistula. Most cases of PWS are sporadic; however some few cases are caused by mutations in the RASA1 gene. Here we report a case of congenital PWS of the left upper limb in a low income country.

Introduction

Parkes Weber syndrome (PWS) is a congenital vascular malformation (CVM) described for the first time in 1907 by Frederick Parkes Weber [1]. It is caracterised by the presence of a capillary malformation, venous malformation, arteriovenous malformation, lymphatic malformation and bone or soft tissue hypertrophy [2]. It affects mostly the lower limb. Although PWS is a distinctive entity, it is still frequently misdiagnosed as Klippel-Trenaunay syndrome which is a triad of capillary malformation, venous malformation, and lymphatic malformation [3,4]. This syndrome had almost similar characteristics as Klippel Trenaunay syndrome, but the presence of an arteriovenous fistula (AVF) [5] and the genetic locus responsible for the onset of the disease made it different and was then called Klippel-Trenaunay-Weber syndrome and later Parkes Weber syndrome [6]. Most cases of PWS are sporadic, however some few cases are caused by mutations in the RASA1 gene and these forms are inherited from an affected parent and follow an autosomal dominant pattern [7]. Most cases of PWS have been reported in high income countries. Here we present a case of Upper limb congenital PWS diagnosed at birth in a low resource setting.

Patient and observation

We received a 1-day-old male infant in the neonatology unit for evaluation of his left hand and forearm swelling with a purplish color and dilated veins at birth. He was born by normal spontaneous vaginal delivery at 38 + 4 weeks of gestation. His mother was 37-year-old and her obstetric history was gravida 3, para 3, living 3. The mother´s pregnancy was well followed without complications. At birth, his Apgar score was 6 and 9 at the first and fifth minute respectively. His birth weight was 3550g (75^{th} to 90^{th} percentile), height was 50 cm (75^{th} to 90^{th} percentile), and the head circumference was 36 cm (above 90^{th} percentile). His vital signs were stable, heart rate 144bpm, respiratory rate 40cpm, SpO2 94% and temperature 37.2°C. The physical appearances are shown in Figure 1, Figure 2 and Figure 3. He presented with hypertrophy of the left upper extremity. There was swelling of the left hand and forearm, venous ulceration on left hand and in comparison with the right upper limb, there was no difference in length but there was 1.4 cm difference in forearm circumference (Figure 1). On palpation there was an enlargement of the soft tissues, swelling of the superficial veins (varicose veins) (Figure 2) and the presence of a thrill. Large capillaries vessels on the left hand were also noted (Figure 3) and the presence of vascular murmurs in the left palm and in the lower-internal one third of the forearm suggesting an arteriovenous

malformation. In addition, no malformation of the fingers was noted. The movements of both arms were free. Cardiac examination was normal as well as renal and spinal ultrasound. The diagnosis of Parkes Weber syndrome was made and Doppler´s ultrasound was requested. Doppler ultrasound of the left upper limb reveals an arteriovenous fistula involving the brachial artery and the cephalic vein at the level of the left elbow (Figure 4). There were also a supernumerary superficial veins with sometimes laminar (venous) flow, sometimes mixed with thrill in certain places, the peak sometimes exceeding 120 cm/s and moreover. These anomalies confirmed the diagnosis of PWS. We explained the pathology to the family and referred the patient to the Yaounde Teaching Hospital (YTH) for multidisciplinary care. The disease was not present in the family.

Discussion

Symptoms and diagnosis: diagnosis of PWS can be made on clinical findings, but the morphology and functional description is important for the management and prognosis of the disease [3]. PWS affect the lower limb in 87.5 % of cases and the upper limb only in 12.5 % of cases as in our patient [3]. It is associated with thromboembolic events, pulmonary venous varicosities, and pulmonary lymphatic obstruction. It can be sometime associated with hydronephrosis [8]. High output cardiac failure in PWS is reported in 31% of cases and it is usually due to large arteriovenous malformation and anemia. This cardiac failure can be fatal in children [3]. Varicose veins can affect both superficial and deep veins and can lead to thrombophlebitis as well as venous ulcers as it is the case in our patient [3]. Therefore, imaging techniques are valuables to evaluate arteriovenous lesions for a better treatment strategy. Usually The gold standard is contrast arteriography but for the precise separation of high-flow and low-flow vascular malformations, digital subtraction angiography (DSA) is the best option [2,3]. Magnetic resonance angiogram (MRA) and magnetic resonance imaging (MRI) are the best option to check for anatomical malformations [9,10].

Treatment and prognosis: the goal of PWS treatment is to improve quality of life and it depends on the patient's age and clinical condition. Conventional sclerotherapy is sometime used as a palliative treatment for PWS, because they reduce many vascular abnormalities in the limbs. It offers good prognosis when used to treat small vascular malformations, and it´s usually indicated for preoperative support as it reduce the size of lesions before surgery or for postoperative therapy [11]. Elastic compression stockings are sometimes useful when there is lymphedema or chronic venous insufficiency (CVI). It prevents deep vein thrombosis and recurrent cellulitis. Furthermore, it provides good protection for traumatic events [12]. Laser therapy can be used as treatment for capillary malformations and it could provide an improved cosmetic appearance [13]. However, depending on the level and type of the lesion, other modalities of treatment like embolization, stent graft implantation, AVM resection and amputation can be used [2,3,9].

Conclusion

PWS is a rare congenital vascular malformation under diagnosed and no cases have yet been reported in low income countries. Dangerous complications can occur in these patients like cardiac failure and the management is multidisciplinary.

Competing interests

The authors declare no competing interests.

Authors' contributions

FDT: conceptualization, project administration, supervision, writing original draft, writing, review and editing. ANM, SPT, NAT, GLN, SN: writing original draft, writing, review and editing. All the

authors have read and agreed to the final manuscript.

Figures

Figure 1: hypertrophy of the upper left limb

Figure 2: swelling of superficial veins

Figure 3: large capillaries vessels on the left-hand

Figure 4: arteriovenous fistula involving the brachial artery and the cephalic vein

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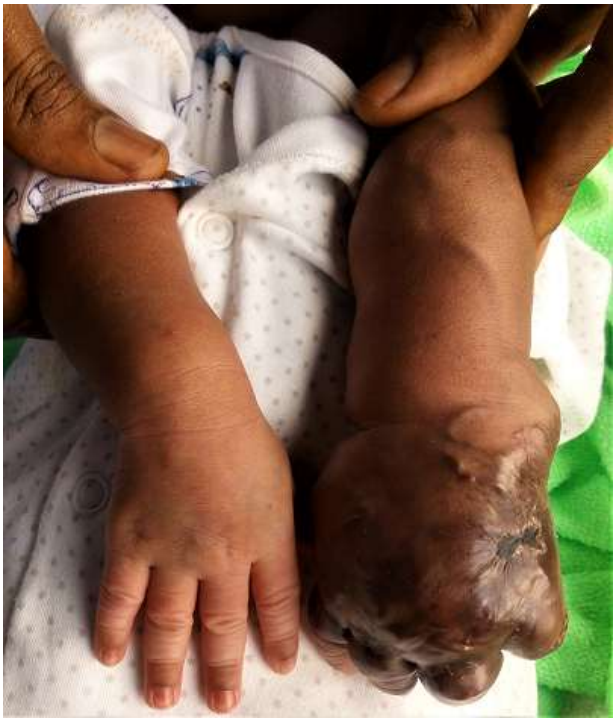

Figure 1: hypertrophy of the upper left limb

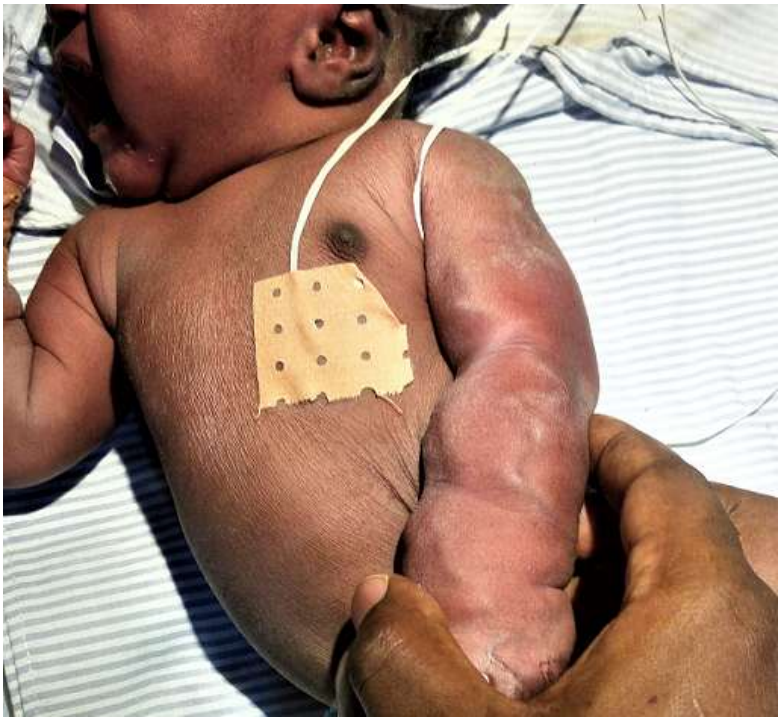

Figure 2: swelling of superficial veins

Figure 3: large capillaries vessels on the left-hand

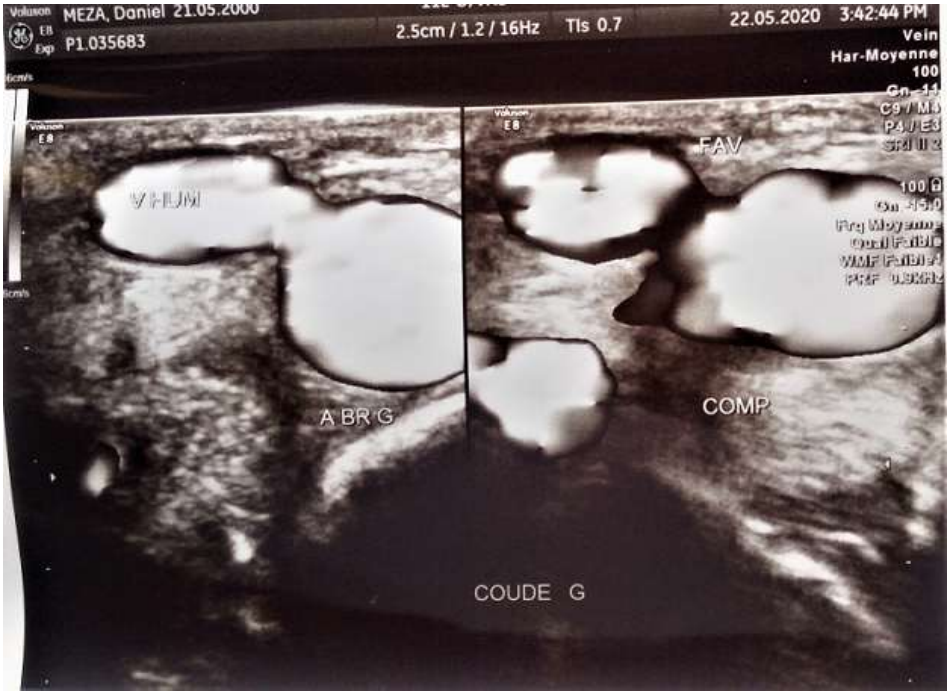

Figure 4: arteriovenous fistula involving the brachial artery and the cephalic vein