

PARKES WEBER SYNDROME: A Rare Vascular Malformation-Imaging And The Usefulness Of Intravascular Intervention

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Abstract

Parkes Weber syndrome is a rare complex, combined capillary and high flow arteriovenous malformation with affectation of an entire limb. Hypertrophy of the affected limb and port wine skin discoloration are commonly associated. This is a case of Parkes Weber syndrome that was first diagnosed at age 5 months as Klippel-Traunanay syndrome but follow up imaging as the child got older revealed a diagnostic feature consistent with Parkes Weber syndrome. Also described are the Magnetic resonance/Magnetic resonance angiography and digital subtraction angiographic features and intravascular management by combined repeated percutaneous embolization, sclerotherapy and coiling.

CASE REPORT

A 5 year old male was brought to the hospital on account of right lower limb enlargement. The parents noticed enlargement and areas of skin discoloration on the right leg since birth. The right lower limb was noticed to be growing disproportionately larger than the left. The pregnancy period was uneventful and no history existed of similar lesions in the family.

Physical examination revealed a conscious active boy, not febrile, pale or icteric, with stable vital signs. Essential systemic findings were in the musculoskeletal system which revealed a disproportionately hypertrophied right lower limb. It measured 11.70cm and 7.18cm at the thighs and 6.23cm and 5.17cm at the legs on the right and left respectively. The boy had a slightly lengthened right lower limb (25.39cm) when compared to the left (22.14cm) at the age of 5 months. The leg was warmer than the contralateral side. A geographic pink skin discolouration was noted over the lateral aspect of the distal leg and dorsum of the foot. He was commenced on conservative management.

The limb was getting increasingly bigger despite conservative management with compression socks and had skin ulceration of the medial aspect of the ankle of the same right lower limb 2 months before any interventional

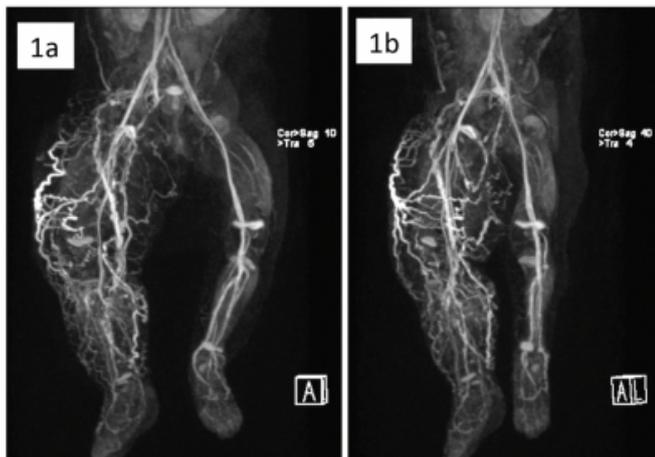
procedures were done. Subsequent examination at age of 5 years still showed right lower limb hypertrophy but to a lesser degree (measurement at the thigh, 12.03cm and 9.95cm, on the right and left respectively). Additionally, a scar was noted in the region of the medial malleolus from a healed ulcer. Other systems were essentially normal. Laboratory investigations; full blood count, electrolyte and urea as well as liver function test were essentially normal.

Diagnostic imaging

Magnetic resonance imaging and magnetic resonance angiography were done at 5 months and 5 years of age. MRI images were acquired by 1.5 Tesla Philips MRI scanner in axial, coronal, and sagittal T2-tirm, axial T1-Vibe Fat-Sat, coronal T1-3D Flash dynamic post Gadolinium contrast (Dataram) administration, and digital subtraction and MIP reconstruction were done. These images show significant hypertrophy of the right lower extremity as evidenced by increase in width and length (Fig 1a & 1b).

Figure 1a&b

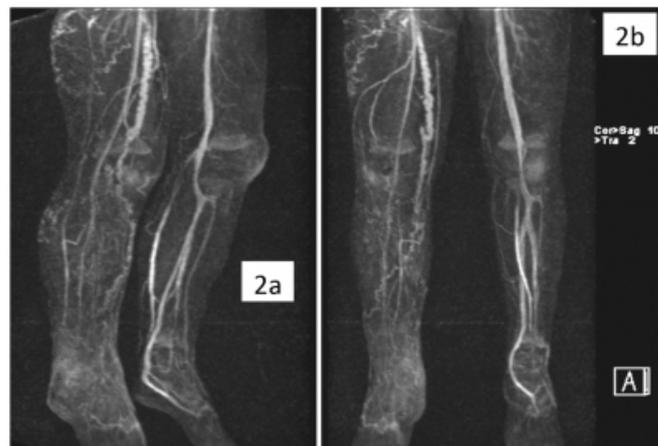
Multiplanar reconstruction of MRI angiography of both lower limbs. Anteroposterior(a) and left anterior oblique view(b) showing the arterial and early filling of multiple tortuous dilated veins involving the subcutaneous tissue and muscles of the leg. Marked Hypertrophy and increased length of the right lower limb noted.



There was an associated increase in the thickness of the subcutaneous tissue in the entire right lower limb. Moderate T2 hyperintense signal alterations were noted in this limb. The soleus and gastrocnemius muscles as well as the soft tissue also show diffuse hyperintense T2 signal alterations. No circumscribed cystic changes were seen. Arterial phase of the MRA showed early filling of multiple extensive varicose venous vessels with a large varix in the medial aspect of the thigh. These abnormal dilated veins were mostly in the subcutaneous and to a lesser extent in the intramuscular compartment (Fig 1a & 1b) of the right lower limb. The arteries of the right leg were normal, the arteries and veins of the left lower limb showed no anomaly. A diagnosis of Klippel-Traunanay syndrome was made based on the imaging findings. Subsequent MRIs at age 5, confirmed no change in above findings; the multiple extensive dilated veins were noted to be from the superficial system while the deep venous system was faintly depicted suggesting hypotrophy (Fig. 2 and 4). However, pre procedure digital subtraction angiography performed to determine the extent and flow characteristics of the vascular lesion showed a large arteriovenous (AV) fistula arising from the common femoral artery and communicating with the femoral vein and the great saphenous vein. Associated flow reversal was also noted, in keeping with Parkes Weber syndrome.

Figure 2a&b

MPR images of MRA of both lower limbs in left anterior oblique(2a)and antero-posterior view (2b) showing Early filling of multiple, tortuous dilated superficial veins in the right lower limb. Left lower limb vessels are normal. Hypertrophy and increased length of the right lower limb still noted.



INTERVENTIONAL THERAPY

The patient had a first embolization and sclerotherapy at age of 5. Embolization of the abnormal veins of the thigh was carried out with glubran, an acrylic intravascular glue agent with lipiodol for opacification. The large varix, originating in the distal third of the right thigh medially was also accessed with a Venflon intravenous cannula and sclerotherapy was performed with a total of 3.5mg of Aethoxysklerol (a sclerosant) mixed with Iopamiro and about 5ml CO₂ gas with satisfactory result (Fig 3).

Figure 3a&b

Digital subtraction angiography images of the Rt lower limb showing multiple abnormal tortuous veins

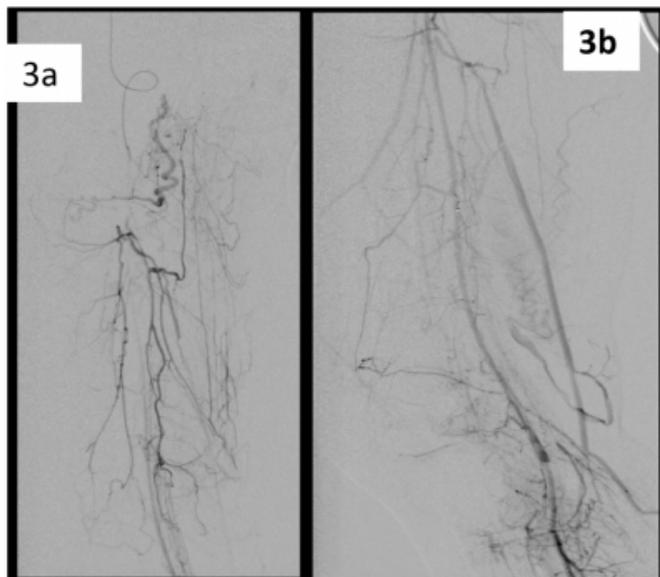


Figure 3c&d

Digital subtraction angiography images of the Rt lower limb. Image 3c shows a large arteriovenous feeder from the origin of the deep femoral artery. Image 3d - DSA image Post embolization and sclerotherapy showing non opacification of the previously noted abnormal veins in 3 a& b

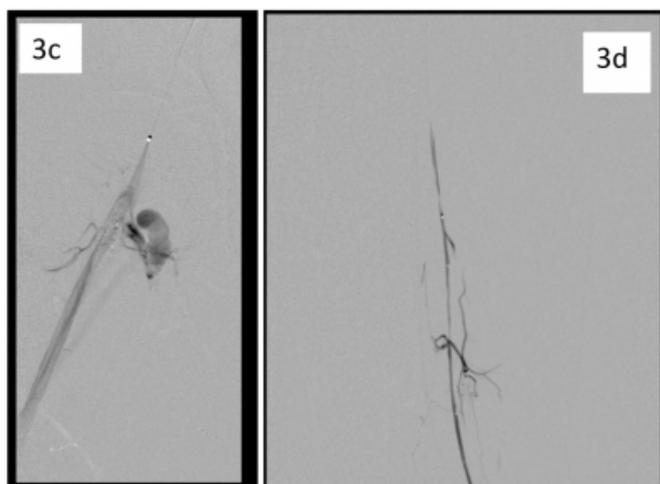
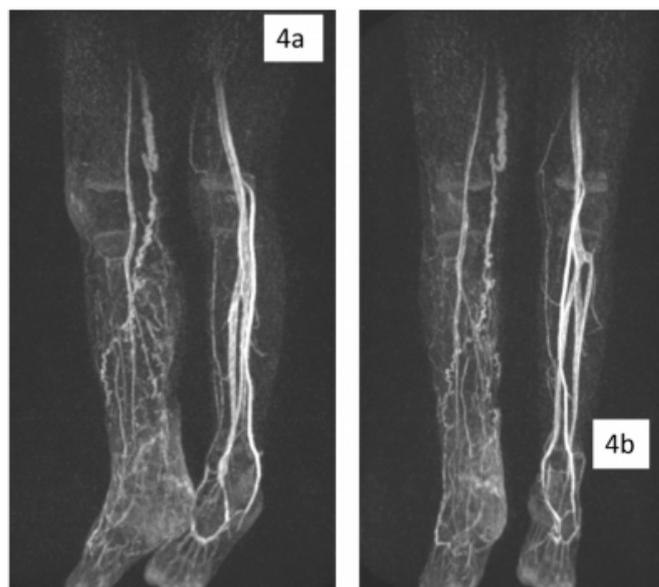


Figure 4a&b

MPR images of MRA (pre 2nd sitting intervention) of both lower limbs in right anterior oblique(a)and anterior posterior view(b) showing Early filling of multiple, tortuous dilated superficial veins involving the subcutaneous and muscle plane of the leg. Similar changes but limited to the subcutaneous tissue is also seen in the right thigh limb. Left lower limb vessels are normal. Hypertrophy and increased length of the right lower limb still noted.



Pre-procedure MRI/MRA prior to the second intervention essentially showed unchanged superficial veins with significant varicose changes medially on the lower leg. However, the multiple abnormal venous blood vessels on the medial side of the proximal femur to the knee were obliterated. Endovascular embolization of the known AV fistula feeder which arises from the CFA at the origin of the Deep femoral artery was done using Guglielmi detachable coils (GDC) (Fig 5).

Figure 5a&b

DSA of Rt lower leg pre coiling (a & b) of the large arteriovenous fistula feeder.

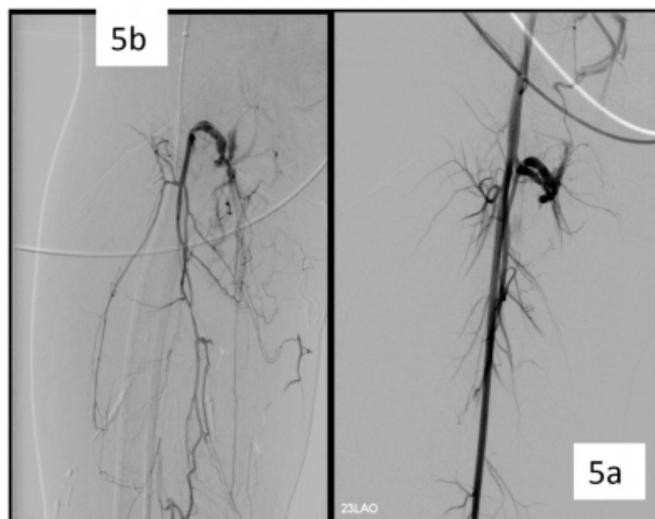
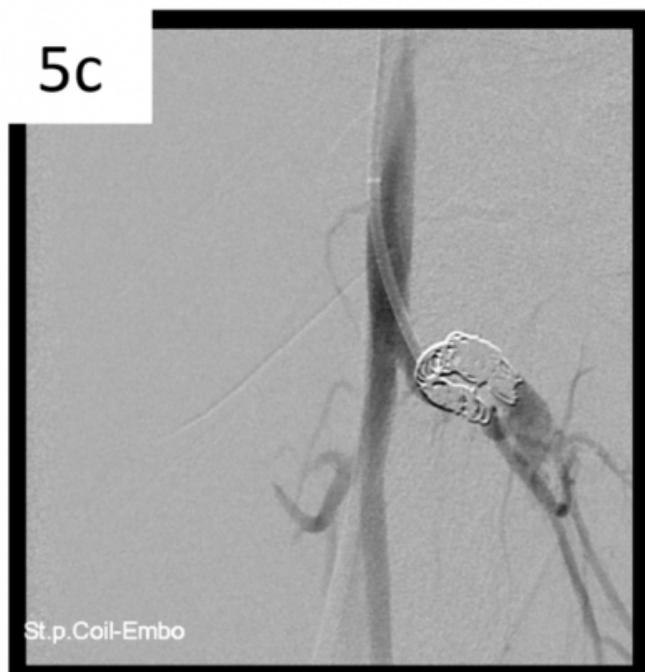


Figure 5c

DSA of Rt lower leg post-coiling embolization of the large arteriovenous fistula feeder



DISCUSSION

Parkes Weber syndrome is a rare congenital complex vascular malformation involving arteries, capillaries and veins with an underlying high flow arteriovenous fistulas (AVF) devoid of capillaries, as well as soft tissue and skeletal hypertrophy of the affected limb [1, 2]. PWS was

first reported in 1907 by and named after Frederick Parkes Weber [1]. No sex predilection is noted in literature [2]. Association with mutation of RAS1 gene coding for p120-RasGTPase-activating protein essential for normal vascular formation have been reported to be responsible for familial expression of some cases of PWS [2, 3, 4] which is inherited in autosomal dominant fashion [5]. There were no familial expressions of similar anomaly in this case.

It is a distinct entity and clearly different from the commonly stated differential diagnosis of Klippel-Traunanay syndrome (K-PS) first described by Maurice Klippel and Paul Trenaunay in 1900 [6, 7] as naevus vasculosus osteohypertrophicus [7]. And also a congenital disorder characterized by port wine stain, abnormal venous structure including venous malformation, varicose veins, soft tissue and osseous hypertrophy [8, 9, 10, 11]

PWS must however be differentiated from K-PS, as the presence of high flow arteriovenous fistula in PWS affects the disease progression, complications and likely choice of treatment modality. While K-PS is a combined capillary lymphatic and venous malformation it is a slow flow anomaly. PWS also a combined capillary arteriovenous malformation, but a fast flowing AVF vascular anomaly [12] and shunting of blood into this lesion could lead to high output heart failure [4].

Cohen in 2000 [8] described the differences between Klippel-Trenaunay syndrome and Parkes Weber syndrome as shown below:

Table 1

	Klippel-Trenaunay syndrome	Parkes Weber syndrome
Types of vascular malformations	Slow flow; capillary, lymphatic, venous	Fast flow; capillary; arterial; venous
Color of cutaneous malformations	Bluish-to-purple.	Pink and diffuse
Arteriovenous fistulas	Insignificant and physiologic.	Very significant
Lateral venous anomaly	Very common.	Not found
Lymphatic malformations	Present	Very rare
Lymphatic vesicles	Present.	Not found
Venous flares	Present.	Not found
Limb affected		
Upper.	5 %.	23%
Lower Limb.	95%.	77%
Limb enlargement	Usually disproportionate, involving soft tissue and bone; macrodactyly, particularly of toes, is common	Arm or leg length discrepancy
Prognosis	Usually good; pulmonary embolism is encountered in about 10% of children.	More problematic, particularly in those who develop heart failure resulting in cardiac enlargement and cutaneous ischemia, requiring limb amputation

PWS affects the lower limbs mostly when compared with the upper limbs [4, 10]. The affected limb is hypertrophied with port wine stain. It is usually warmer with a bruit or thrill due the arteriovenous fistula and usually longer than the contralateral limb. The vascular malformation gets larger as the child grows and is associated with vessel dilatation, obstruction and thrombosis [13], cardiac enlargement [10, 14] or high output heart failure [4]. Rare case of associated shorter limb has also been reported in the literature [2]. This case showed affection of the right lower limb, significant hypertrophy of the soft tissues (subcutaneous tissue and the muscles) and the bones with limb length discrepancy with associated port wine cutaneous stain. No cardiac enlargement or failure was however seen in this case.

Imaging by magnetic resonance (MR) and imaging with MR angiography is the modality of choice in children. Features on MRI include hypertrophy of muscles and bones of affected limb, abnormal signal intensity and contrast enhancement and multiple arterial and venous dilatations [15, 16]. Digital subtraction angiography is done prior to intravascular therapy. Other modalities are vascular ultrasound, though cheap but operator dependent, and Computed Tomography in the adult cases. Diagnostic digital subtraction angiography in this case, also revealed a fast flow AVF, consistent with PWS. The diagnosis of Klippel Trenaunay syndrome at infancy, without angiographic evaluation, may be incorrect. We suggest that angiographic examination, as the child grows, to evaluate the presence or absence of AVF, a crucial diagnostic feature of PWS.

Treatment depends on presence of symptoms and its severity [17]. Nonoperative management include suitable foot wear, graduated compression stockings, and garments. Non operative management was instituted initially in the case presented. Operative management is indicated for varicose veins and leg length discrepancy of more than 2cm [2]. PWS requires repeated surgical surgery or interventional therapy. The therapy of choice is repeated embolization. But limb amputation may be resorted to in cases of resistant progressive disease complicated by uncontrollable massive bleeding due to ulcerations, necrosis or cardiac enlargement [17]. Conservative management was commenced initially in this case. The skin ulcerations and detection of a fast flow AVF necessitated treatment with repeated sclerotherapy and embolization with liquid agent and coiling of the high flow fistulae.

CONCLUSION

A case of Parkes Weber Syndrome, a rare complex vascular malformation with characteristic progressive limb hypertrophy and length increase first diagnosed as Klippel-Traunay Syndrome is presented. An arteriovenous fistula, a prominent feature of PWS, was later discovered on imaging as the child grew. This case highlights the clinical progression, complication, the need for follow up imaging of a diagnosed Klippel-Traunay syndrome at infancy. The diagnosis of Klippel Trenaunay syndrome at infancy without angiographic evaluation may be incorrect. We suggest that angiographic examination as the child grows to evaluate the presence or absence of AVF. This is crucial not only to distinguish PWS from K-TS but also to inform the managing physician about possible complications and management options.

This case study also emphasize the critical role of radiological interventional treatment with combination of repeated sclerotherapy, embolization with liquid agents and coils in the management of PWS.

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