Parkes Weber's Syndrome

Nomenclature

The terms Klippel-Trénaunay-Weber syndrome and Klippel-Trénaunay-Parkes Weber syndrome have sometimes been used synonymously with Parkes Weber's syndrome (PWS). However, current usage favours the term Parkes Weber's syndrome. Note that Klippel-Trénaunay syndrome (without the 'Parkes' or 'Weber') is a separate condition (limb overgrowth and a slow-flowing vascular malformation without significant AVFs - clinically different from PWS). [1, 2]

Definition [3, 4]

Parkes Weber's syndrome (PWS) is vascular malformation comprising:

- A capillary arteriovenous malformation, manifest as a visible red skin lesion.
- Arteriovenous fistulae (AVFs).
- Lymphoedema.
- Overgrowth of a limb.

PWS is classified as a complex combined vascular malformation.

Epidemiology and aetiology

- Parkes Weber's syndrome (PWS) is rare. [5]
- Most cases are sporadic, although familial cases have been reported. [3, 6]
- A recent study suggests that PWS (and other capillary malformation-arteriovenous malformations) may be linked to mutations of the RASA1 gene. [7, 8]

Presentation [3, 4, 9, 10]

Onset:

- May present antenatally (on ultrasound), at birth, or may develop during childhood.

Clinical features in the affected limb:

- A congenital, red or pink skin lesion (a 'geographical' red stain), which is a high-flow lesion. [11]
- Limb enlargement - including muscle and bone hypertrophy, with an increase in limb length and girth. One case involving a shortened limb is reported. [12]
- Signs of a vascular shunt in the affected limb, eg warmth; dilated veins; a thrill, bruit or pulsation.
- Lymphoedema - localised or diffuse. Lymphatic vesicles may be visible in the skin.
- May have limb pain, due to vessel enlargement.
- In some cases, the skin lesions may bleed easily, eg on minor trauma. [11]
Distal skin changes in the limb (due to distal vascular steal), eg ischaemic ulcers, pigmentation and fibrosis.

**Diagnosis and investigations**[^3]

The diagnosis can usually be made clinically, without the need for imaging. A bedside audible Doppler ultrasound can confirm vascular shunting.

Various imaging methods can be used to assess the extent of lesions:

- Plain X-rays - show lytic bone lesions and limb-length discrepancy.
- MRI scans - can show enlarged limb muscles and bones, the extent of the vascular lesion, and the high-flow nature of the vascular malformations.[9, 13, 14]
- Catheter angiography is used in some cases.[15]

**Differential diagnosis**

- Klippel-Trénaunay syndrome (slow-flow capillary lesions without significant arteriovenous fistulae (AVFs)).
- Other vascular malformations, eg port-wine stains[^16] (tend to be purplish in colour rather than the pink-red stain found in Parkes Weber's syndrome (PWS)).
- Other causes of lymphoedema.

**Management**[^3]

Conservative management is preferred, if possible, since invasive treatment may worsen the arteriovenous fistulae (AVFs). Multidisciplinary care is often appropriate.

**Conservative treatments:**

- Prevention of trauma (lifestyle modification, eg care with sporting activity) - since trauma may worsen the AVFs.
- First aid advice for patients if they have lesions prone to bleeding - apply firm pressure and seek medical help.
- Elastic hosiery to reduce lymphoedema and vascular steal.
- Avoid laser treatment of the skin lesions - this can worsen the shunting through AVFs.

**Orthopaedic care for the limb-length discrepancy:**[^4]

- Monitor limb growth.
- Treatment is conservative if possible.
- Stapling epiphysiodesis (eg of the knee cartilages) may be performed to limit leg length, but the procedure may worsen the arterial venous malformation in the limb.

**Vascular treatment:**[^17] This may be required if there are troublesome complications such as pain, ulceration or cardiac failure. Possible treatments are:

- Arterial embolisation (but this procedure often fails to control the shunting in Parkes Weber's syndrome (PWS)).[^3, 18, 19]
- Surgical resection of the lesion may be possible in some cases.
- Limb amputation may be required in some cases.[20]

**Complications**[^3]
Skin:
- Cosmetic problems with the appearance of the lesions.
- Some skin lesions may bleed easily, eg on minor trauma - patients need advice about first aid.
- Ischaemic ulcers distal to the lesion.
- Recurrent skin infections due to lymphoedema.

Cardiovascular:
- Limb pain resulting from vessel dilatation.\[10\]
- High-output cardiac failure - due to the high-flow shunting lesions.\[5, 15\]
- One case report describes disseminated intravascular coagulation following a leg fracture in a patient with Parkes Weber’s syndrome (PWS).\[21\]

Orthopaedic:
- Pelvic tilt and scoliosis due to leg-length discrepancy.
- Pathological fractures due to lytic bony lesions.\[10\]

Prognosis\[1\]
The deformity tends to progress with time; the affected limb continues to show increased growth until epiphyseal closure.

Further reading & references
- Photograph of Parkes-Weber syndrome, Birthmarks US website, accessed April 2011

8. Parkes Weber Syndrome, Online Mendelian Inheritance in Man (OMM)
11. Capillary vascular malformation, DermNet NZ, 2004