Parkes Weber's Syndrome

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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).

Definition

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.
- Most cases are sporadic, although familial cases have been reported.
- A recent study suggests that PWS (and other capillary malformation-arteriovenous malformations) may be linked to mutations of the RASA1 gene.

Presentation

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.
- Limb enlargement - including muscle and bone hypertrophy, with an increase in limb length and girth. One case involving a shortened limb is reported.
- 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.
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 (OMIM)
11. Capillary vascular malformation, DermNet NZ, 2004