CASE REPORT

Challenges in orthopaedic management of Parkes-Weber syndrome

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SUMMARY

A 16-year-old boy with a diagnosis of Parkes-Weber syndrome presented with a lower leg discrepancy of 3 cm for orthopaedic management. He had the triad of red skin lesion, lymphoedema and overgrowth of the right leg and multiple arteriovenous fistulae confirmed by angiography. Considering the risk of aggravating the vascular lesion, we decided conservative management of unequal limb lengths as long as this is well tolerated.

BACKGROUND

This is a case of a rare complex combined vascular malformation associated with accelerated growth of affected limb that represents the difficulties in orthopaedic management of such situations.

CASE PRESENTATION

We report a 16-year-old boy, born and resident in the Democratic Republic of São Tomé and Príncipe. He had a medical history of malaria (at 6 years), glucose dehydrogenase 6 phosphate deficit, ferropepic anaemia, hepatitis C virus chronic infection and nocturnal enuresis. His grand-uncle had hypertrophy of a lower limb.

At 9 years he was evacuated to Portugal for investigation of the lower limb asymmetry with hypertrophy of the right lower limb and recurrent cutaneous infections with systemic signs and need for hospitalisation. He presented red cutaneous staining, lymphoedema with hypertrophy of girth and increased limb length, trophic cutaneous lesions and inflammatory signs of the right leg. He was treated for erysipela with antibiotics with a good response. Doppler ultrasound showed no alterations and vascular surgery evaluation suggested Klippel-Trénaunay-Weber syndrome.

He returned to São Tomé and Príncipe, where he was treated prophylactically with monthly penicillin, with diminished frequency of infections.

At 11 years he was re-evaluated in vascular surgery consultation at our centre due to persistence of limb asymmetry. Soft tissue hypertrophy, varicosities and lymphatic vesicles were also apparent. An angiography was performed, showing multiple arteriovenous fistulae, not evident in the previous evaluation by Doppler ultrasound. Lymphoscintigraphy of the lower limbs showed a permeable lymphatic system, normal lymph drainage and lymphoedema involving right foot and distal half of the leg. He also performed a lower limb MRI; the fat-saturated T2 image showed dilated vascular structures in the right lower limb involving subcutaneous and multiple muscle compartments, flow voids consistent with

arteriovenous malformation and subcutaneous oedema (figure 1).

The clinical characteristics (vascular malformations and soft tissue hypertrophy) and the presence of arteriovenous fistulae confirmed the diagnosis of Parkes-Weber Syndrome (PWS).

He returned to his country, with no maintenance of regular monitoring by vascular surgery or orthopaedics teams.

INVESTIGATIONS

Owing to the possible coexistence of vascular malformations, namely haemangiomas, of the abdomen, pelvis, kidney and, more rarely, of the central nervous system, urine exam, abdominal, renal and bladder ultrasound and cranial CT scan were performed, which were normal.

Cardiac enlargement, possibly with cardiac insufficiency, can also be found. However, in this patient, chest radiography and echocardiogram showed no alterations.

DIFFERENTIAL DIAGNOSIS

PWS and Klippel-Trénaunay Syndrome are rare complex combined vascular malformations that consist of a triad of: capillary angioma of a limb, venous malformation and hypertrophy of soft tissue and/or bone. They are distinguished by the presence of apparent arteriovenous fistulae in PWS.^{1 2} It is important to differentiate between the two syndromes because PWS has a worse prognosis for limb viability.

OUTCOME AND FOLLOW-UP

At 16 years, he was evaluated in Paediatric Orthopaedic consultation at our centre for leg length discrepancy correction. He maintained circumferential and longitudinal hypertrophy of the right lower limb involving muscles and bone (figure 2), with a discrepancy of 3 cm (figure 3).

We decided conservative management as long as limb discrepancy is tolerated. We prescribed a knee insertion to minimise limb length difference and asked to follow-up.

DISCUSSION

The PWS presents at birth or during early infancy with a red or pink skin lesion, with other changes arising during childhood: lymphoedema, overgrowth of a limb, a pulsatile mass with thrill or bruit, trophic skin lesions and limb pain. Most cases are sporadic although familial cases have been reported with autosomal dominant pattern of inheritance and variable expressivity. Our patient

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Figure 1 Lower limb MRI showing dilated vascular structures in the right lower limb involving subcutaneous and multiple muscle compartments, flow voids consistent with arteriovenous malformation and subcutaneous oedema.

had a relevant family history and it is possible that this PWS is caused by a genetic mutation. Mutations have been described in RASA1 gene.^{3 4} Although his grand-uncle had lower limb hypertrophy, the specific mutation of RASA1 gene was not researched, because he was not available for study and this result would not interfere with treatment decisions in this patient who meets the diagnostic criteria of PWS.

Diagnosis is essentially clinical and confirmed by imaging. Work-up of the lesion involve mainly non-invasive imaging: Doppler ultrasound that confirms vascular shunting; standard radiographs that may reveal lytic bone lesions and limb length discrepancy; an MRI that shows the extent of vascular lesion, enlarged muscles and bones and high flow nature of the vascular malformation.^{5 6}

The vascular malformation in PWS usually worsens with time. Possible complications are recurrent skin infections due to lymphoedema, limb pain, ulceration, haemorrhage after minor trauma, high output cardiac failure, pelvic tilt and scoliosis,

pathological fractures and degenerative joint disease at an early age. There have been described brain abnormalities such as haemorrhage, infarction, vascular malformations, hydrocephalus, atrophy and calcifications.⁵ Conservative and symptomatic treatment include pain management, prevention of trauma, bleeding control, compression garments and heel insertions for discrepancies of 1.5 cm or less. In some cases may be necessary invasive treatment in a multidisciplinary approach to venous control and orthopaedic management of unequal limb lengths.^{6 7}

Orthopaedic management of unequal limb lengths in vascular bone syndromes is difficult to decide. In PWS leg length discrepancy may reach 10 cm, with possible complications of pelvic tilting and scoliosis. To avoid such a course, stapling epiphysiodesis of the knee cartilages is often performed, but this orthopaedic procedure may augment the worsening of the arterial venous malformation in the limb,⁷ thus, we considered left tibia elongation for surgical correction.

Figure 2 Hypertrophy and trophic cutaneous lesions of the right lower limb.

Figure 3 Hypertrophy of muscles and bones of the right lower limb, with discrepancy of 3 cm.

The prognosis of this patient depends on the local infections and trauma, limb length discrepancy at the end of growth and the morbidity of the treatment procedure chosen.

Learning points

- ▶ The presence of arteriovenous fistulae distinguishes Parkes-Weber syndrome from the other clinically similar entity Klippel-Trénaunay Syndrome.
- ▶ The preferable approach in the treatment of unequal limb length is conservative.
- ▶ Epiphysiodesis of the knee cartilages in the affected limb was associated with aggravating the worsening of vascular malformation. Elongation of contralateral limb is a possible alternative.

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