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Re: Parkes Weber Syndrome: a case of right lower limb hypertrophy

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Dear Editor,

The Images for Surgeons article 'Parkes Weber syndrome: a case of right lower limb hypertrophy' by Sreedharan et al.¹ describes a patient diagnosed as Klippel-Trenaunay syndrome, who the authors considered to have Parkes Weber syndrome. Parkes Weber is caused by a germline mutation in the RASA1 gene and Klippel-Trenaunay is one of a spectrum of conditions due to somatic mosaic mutations in the PIK3CA gene, collectively called PI3Kinase Related Overgrowth Spectrum (PROS)². Cases like this illustrate the importance of correct diagnosis of these conditions.

We have recently treated a remarkably similar patient, originally thought to have Klippel-Trenaunay but on referral to another hospital with a leg ulcer was diagnosed with Parkes Weber and high output heart failure and treated by embolization. This was followed by worsening of the leg ulceration, secondary infection, and two failed attempts at skin grafting. When transferred to our care the leg was ischaemic with extensive ulceration. Amputation was the only viable option. Ultrasound examination and review of the imaging suggested that rather than Parkes Weber, a PROS condition was a more likely diagnosis. Although hypoplasia of axial veins can occur in Klippel-Trenaunay, dilation of persistent embryonic veins and of axial veins, as in this case, is also possible. Such pathologically enlarged veins create a risk for pulmonary embolism. A lung scan confirmed the presence of multiple pulmonary emboli which we concluded was the cause of her persistent shortness of breath. The size of the IVC precluded the use of a standard filter, so a 'bird's nest' filter was inserted into the iliac vein, before the patient underwent amputation through the knee. She was eventually discharged from hospital some six months after her original admission for a leg ulcer.

High output heart failure is described in cases of AVM, but is not common, particularly in Parkes Weber syndrome. Arteriovenous shunts in Parkes Weber are small and diffuse throughout the limb and so poorly suited to embolic treatment³. They are not generally visible on CT angiography. Attempted embolization can have disastrous results as it did in this case. In contrast, pulmonary embolism is a relatively common complication of lesions in the PROS spectrum, including Klippel-Trenaunay syndrome, and repeated subclinical episodes over long periods can result in pulmonary hypertension which may be mistaken for high output failure. Diagnosis of complex vascular anomalies is difficult, and patients are best treated by a multidisciplinary team specializing their management. All surgeons, however, should be aware of the risk of pulmonary embolism in patients with low-flow vascular malformations.

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