Misdiagnosis of Klippel-Trenaunay Syndrome

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To the Editor We read with interest the case report, “A 17-year-old Girl with Klippel-Weber Syndrome Complicated with a Pulmonary Thromboembolism and RV Thrombus” (1). In this report, the authors described a patient with “Klippel-Weber syndrome”, pulmonary thromboembolism and a right ventricular thrombus. Thrombectomy was performed and an inferior cava vein filter was placed.

We compliment the authors for their successful management of this challenging thromboembolic event. Nevertheless, we believe that the report contains a few inaccuracies, particularly regarding the diagnosis of Klippel-Trenaunay syndrome (KTS).

1. KTS is characterized by capillary (port-wine stain), lymphatic and venous malformations in an overgrown limb (2). The capillary malformations are typically located on the lateral aspect of the limb and contain lymphatic cutaneous vesicles (3).

2. The authors described the patient’s leg as being “edematous and swollen,” while neither lymphatic components nor overgrowth were noted.

3. The photographs showed extensive port-wine stains on the left lower extremity. The authors erroneously referred to these malformations as “hemangiomas” and “nevi.” Infantile hemangiomas are benign vascular tumors characterized by proliferation and involution phases in early infancy and childhood, while port-wine stains are malformations, not tumors.

4. The authors also described similar lesions on the forehead and perioral region. These are not classic features of KTS.

5. Parkes Weber syndrome is characterized by capillary malformations on overgrown limbs with diffuse hypervascularity and is not merely “a similar condition to KTS with arteriovenous rather than venous malformations,” as the author stated. Klippel-Trenaunay and Parkes Weber syndromes are vastly different clinical entities. Parkes Weber syndrome is caused by a RASA1 mutation in many patients, while KTS is caused by a PIK3CA somatic mutation (4, 5). Therefore, the use of combined eponyms, such as “Klippel-Weber” or “Klippel-Trenaunay-Weber,” is confusing and should be avoided.

6. The focus of the report was the management of the patient’s acute presentation. However, the etiology of the thromboembolism was not clearly elucidated. Acute thromboemboli in patients with KTS and other syndromes with phlebectasia typically originate in the marginal venous system or ectatic pelvic, mesenteric or other veins. To reduce the risk of thromboembolism, the use of endovascular closure of the large anomalous veins is recommended, preferably in early childhood (3).

We believe that the diagnosis of KTS in the presented patient is incorrect. Unfortunately, misdiagnoses in the field of overgrowth syndromes with complex vascular anomalies are frequent, particularly concerning KTS (6). It is superfluous to stress the value of conducting accurate analyses of the clinical and imaging findings of various vascular anomalies and using proper terminology.

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References


