The association of Klippel-Trenaunay syndrome and abdominal aortic aneurysms

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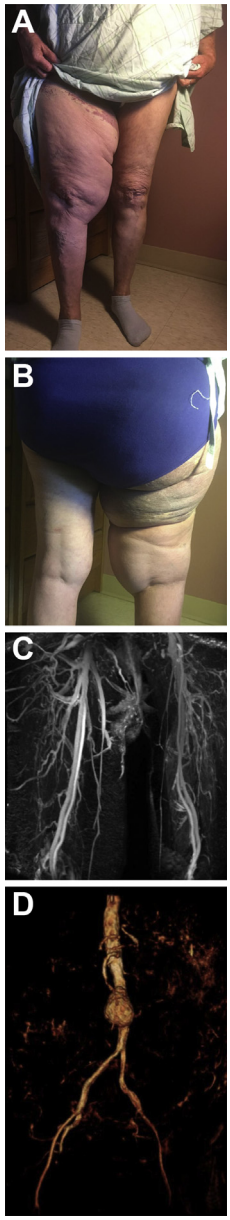
A 71-year-old woman described a gradual progression of right lower extremity swelling, which had been a lifelong problem. Physical examination revealed a right lower extremity femoral port wine stain (*A*), superficial venous varicosities (*A*), right gluteal and posterior thigh soft tissue swelling, and violaceous skin coloration, consistent with venous malformation (*B*). The right leg was longer than the left and the right thigh and calf were significantly larger than the left. There was 2+ right ankle edema and right thigh lymphedema, with only trace edema on the left. Magnetic resonance angiography demonstrated diffuse soft tissue hypertrophy with numerous dilated superficial veins in the right thigh and gluteal region (*C*), without evidence of arteriovenous malformation or venous thrombosis. There was also an infrarenal abdominal aortic aneurysm (AAA) with a maximum dimension of 4.3 cm (*D*/Cover).

Based on clinical and angiographic findings, the patient was diagnosed with Klippel-Trenaunay syndrome (KTS), which is characterized by cutaneous capillary, venous, and lymphatic malformations in an extremity, more commonly the leg, in combination with soft tissue and/or bony hypertrophy, which begin in infancy and are typically unilateral.[1] Magnetic resonance angiography is the diagnostic imaging study of choice. The presence of an arteriovenous malformation excludes the diagnosis of KTS and favors Parkes-Weber syndrome.[1] KTS has been associated with mutations in the *PIK3CA* gene, which encodes a subunit of phosphatidylinositol 3-kinase (PI3K), a regulator of cell growth, migration, and survival. Treatment consists of compression therapy, pulsed dye laser therapy, and excision of venous varicosities and malformations.[2]

This patient was managed successfully with compression therapy. The patient provided consent for publication of her case. Although AAA is not typically considered a component of KTS, two cases with AAA have been reported, one of whom was a teenager.[3,4] Our report further strengthens the association between KTS and AAA. Furthermore, other arterial aneurysms have been reported in KTS, including renal, cervical, carotid, vertebral, and popliteal.[5] Based on this case and the current literature, we propose that all patients with KTS be screened for AAA, and recommend that arterial aneurysms, in addition to venous and lymphatic pathology, be included in the vascular spectrum of KTS.



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Author conflict of interest: none.

Presented at the Missouri American College of Physicians 2018 Chapter Meeting, Osage Beach, Mo, September 13-18, 2018.

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The editors and reviewers of this article have no relevant financial relationships to disclose per the Journal policy that requires reviewers to decline review of any manuscript for which they may have a conflict of interest.

J Vasc Surg Cases and Innovative Techniques 2019;5:343-4
2468-4287

https://doi.org/10.1016/j.jvscit.2019.04.001

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Submitted Mar 11, 2019; accepted Apr 3, 2019.