Case Report

Dermoscopy and trichoscopy in Klippel-Trénaunay syndrome

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Abstract

Klippel-Trénaunay syndrome (KTS) is a slow-flow vascular malformation characterized by the triad of capillary malformation, limb hypertrophy, and venous malformation, with or without lymphatic malformation. Diagnosis requires the presence of at least 2 of these 3 clinical features; however, there is no consensus regarding dermoscopic or trichoscopic findings. We present the case of a 54-year-old woman with a history of congenital arteriovenous malformation who exhibited clinical, histopathologic, dermoscopic, and trichoscopic findings suggestive of KTS associated with a lower right limb ulcer. We describe a case of KTS diagnosed in adulthood and highlights the potential use of adjunctive diagnostic tools, including trichoscopy and dermoscopy.

Introduction

Klippel-Trénaunay syndrome (KTS), according to the 2018 classification of the International Society for the Study of Vascular Anomalies (ISSVA), is defined as a vascular malformation associated with other anomalies and is characterized by capillary and venous malformations and limb hypertrophy, with or without lymphatic malformations. Although its exact incidence and prevalence are unknown, KTS occurs sporadically and shows no predilection for sex or race.[1]

A possible somatic variant in the *PIK3CA* gene has been identified in some familial cases, leading to activation of protein kinase B (AKT) and the mammalian target of rapamycin (mTOR) pathway.[2] This activation promotes cellular proliferation and angiogenesis, stimulating tissue growth and development. KTS may be associated with complications such as coagulopathies, chronic thromboembolism, bleeding, lymphedema, soft tissue infections, and pain.

Diagnosis is primarily clinical and may be complemented by additional studies, such as magnetic resonance imaging (MRI) with and without contrast, to assess disease extent.[3] Currently, noninvasive tools such as dermoscopy are increasingly recognized as useful diagnostic adjuncts. There is no curative treatment for KTS, and management requires a multidisciplinary approach aimed at symptom control and prevention of complications. Therapeutic options include compression therapy, sclerotherapy, and inhibitors of the AKT–mTOR pathway.

Case Synopsis

We present the case of a 54-year-old woman from Bogotá, Colombia, with a history of congenital arteriovenous malformation without prior diagnostic evaluation, who was admitted for an ulcer of the right lower limb with an 8-year history. On physical examination at admission, homogeneous erythematous-violaceous macules with a reticulated pattern were observed, appearing as isolated and confluent lesions interspersed with areas of normal skin, predominantly affecting the right hemibody. There was lymphedema involving the entire right upper limb. On the mid-third of the ipsilateral lower limb, a large ulcer was noted with erythematous, indurated borders, exposure of tendon and muscle fibers, and a base covered with granulation tissue, involving the entire posterior surface and measuring approximately 15 cm in height and 18 cm in width, without secretion (**Figure 1**).

Dermoscopy of lesions on the scalp, dorsum of the hand, and right shoulder revealed an erythematous-violaceous background with lacunar structures resembling frogspawn (**Figure 2**). Biopsy and histopathologic examination of the ulcerated skin demonstrated proliferation of small capillaries interspersed with adjacent cavernous venous spaces (**Figure 3**). Doppler ultrasound of the right lower limb, including venous and arterial studies, was negative for thrombosis and revealed no additional abnormalities. Ophthalmologic evaluation showed bilateral scleral melanosis, hyperpigmented irises with freckling,

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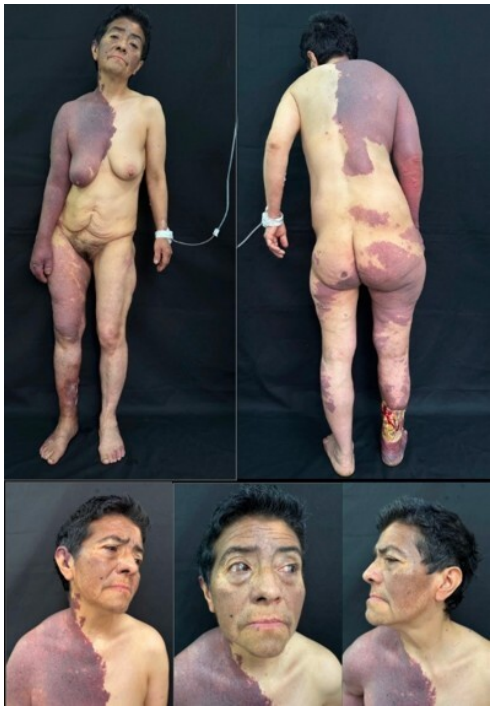

Figure 1. *Clinical presentation of Klippel-Trénaunay syndrome. Erythematous-violaceous macules with a reticulated pattern, appearing as isolated and confluent lesions interspersed with areas of normal skin, predominantly affecting the right hemibody. Marked lymphedema of the right upper limb is present. In the mid-third of the ipsilateral lower limb, a large ulcer with erythematous and indurated borders, exposure of tendon and muscle fibers, and a granulation tissue base is observed, involving the entire posterior surface.*

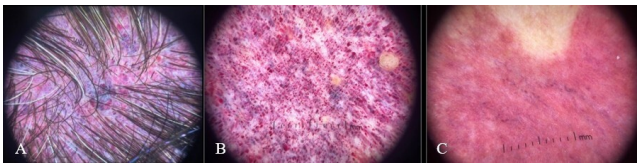

Figure 2. *Dermoscopic findings of vascular lesions on the **(A)** scalp, **(B)** dorsum of the hand, and **(C)** right shoulder showing an erythematous-violaceous background with lacunar structures resembling frogspawn, corresponding to dilated vascular spaces.*

and no evidence of neovascularization in the anterior chamber.

Case Discussion

KTS is defined by the ISSVA as a capillary–venous malformation associated with limb hypertrophy, with or without lymphatic malformation. It is a rare condition, and its exact incidence and prevalence worldwide remain unknown, with no predilection for sex or race.[1] Regarding etiology, most cases occur sporadically; however, familial inheritance has been reported. In many of these patients, a postzygotic somatic variant in the *phosphatidylinositol 4,5-bisphosphate 3-kinase catalytic subunit alpha* (*PIK3CA*) gene has been identified. This gene plays a key role in cell growth and tissue development. Gain-of-function variants in *PIK3CA* lead to activation of the AKT and mTOR pathways, driving 2 major cellular processes: cell proliferation and angiogenesis.[4,5]

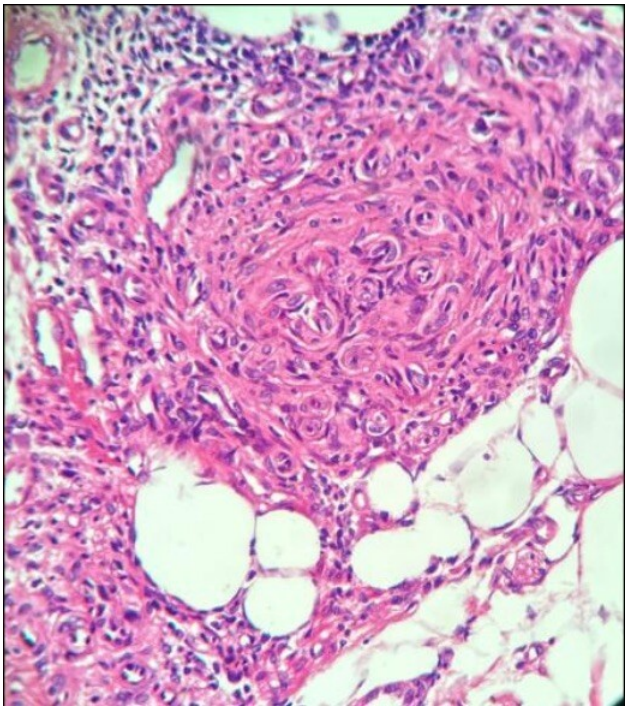

Figure 3. *Histopathologic findings of ulcerated skin demonstrating proliferation of small capillaries interspersed with adjacent cavernous venous spaces, consistent with a slow-flow vascular malformation (hematoxylin-eosin, original magnification ×200).*

The clinical presentation consists of a classic triad. The first component, cutaneous vascular macules, typically appears at birth, most frequently on the lower extremities, with a geographic or segmental distribution. In the present case, the patient exhibited this cutaneous involvement since childhood, affecting the entire right hemibody. The second feature is venous disease, defined by the persistence of embryonic venous structures lacking valves, which predisposes to the development of dilated and tortuous varicosities. These may result in venous insufficiency and complications such as venous ulcers, as observed in our patient. The third component is overgrowth, manifested as increased circumference or length of the affected limb, most commonly involving the lower extremities, with or without alterations in phalangeal shape and size. This was evident in our patient, who presented with right-sided hemihypertrophy, limb length discrepancy, scoliosis, compensatory pelvic tilt, gait disturbance, and pain. Additionally, some patients may exhibit lymphatic malformations, clinically and histopathologically characterized by lymphedema, which was also present in this case.[1-3]

Diagnosis is primarily clinical, based on the presence of the triad, and may be supported by complementary studies such as venous and arterial Doppler ultrasound and MRI (with or without contrast). These modalities help define the extent and nature of vascular and lymphatic abnormalities, as well as bone and connective tissue involvement in patients with KTS.[5]

Noninvasive diagnostic tools such as dermoscopy and trichoscopy have gained increasing relevance in dermatology. However, in KTS, there is currently no consensus regarding specific dermoscopic or trichoscopic features. The literature is scarce, with only 2 reported cases describing dermoscopic findings in KTS and general descriptions of vascular malformations. Reported dermoscopic features include dilated arborizing red vessels, red lagoons on a homogeneous pink background (which may appear brownish upon compression), and white veils surrounding red-purple lagoons. In patients with lymphedema, typical dermoscopic findings include frogspawn-like structures, many of which were observed in our patient (**Figure 2B–C**). To date, no trichoscopic descriptions of KTS have been published, making this case unique (**Figure 2A**).[6-8]

Although histopathologic examination is not required for diagnosis, findings may include ectasia in the superficial dermis, irregular vessels lined by a single layer of endothelial cells, absence of a tunica intima, and limited smooth muscle.[2,3,5] These features were observed in our patient's histopathologic evaluation (**Figure 3**).

There are currently no curative therapies or standardized management guidelines for KTS; therefore, treatment should be multidisciplinary and symptom oriented. In patients with severe disease, targeted therapies such as mTOR inhibitors (sirolimus) and *PIK3CA* inhibitors (alpelisib) have demonstrated improvement in vascular malformation size, hemihypertrophy, and symptoms, and are generally considered safe with few adverse effects.[9-11] Surgical and procedural options include pulsed-dye laser therapy, carbon dioxide ablative laser, sclerotherapy, ligation, phlebectomy, and, in severe cases, complex surgical management by multidisciplinary teams.[1,3] Prognosis depends on the extent of disease and the presence of complications, which contribute significantly to morbidity. The primary causes of mortality include deep venous thrombosis, pulmonary embolism, disseminated intravascular coagulation, and sepsis.[1,3]

Conclusion

This case highlights the importance of recognizing KTS. Although rare, maintaining clinical suspicion and achieving timely diagnosis can have a significant impact on morbidity, mortality, and quality of life, as early management may prevent complications such as those observed in our patient. Furthermore, this case is particularly relevant as it represents one of the few reported cases with dermoscopic findings and, to our knowledge, the first to include trichoscopic evaluation, which may provide useful diagnostic clues for dermatologists assessing patients with suspected KTS.

Potential conflicts of interest

The authors declare no conflicts of interest.

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