Klippel-Trenaunay-Weber Syndrome with Polydactyly: A Case Presentation

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Purpose
Klippel and Trenaunay in 1900 first described a condition featuring a large vascular nevus with dermalatrical distribution, varicosities and limb enlargement. In describing an affected extremity, Parkes-Weber in 1918 described these conditions in combination with arteriovenous fistulas.1 It has been estimated that approximately 25% of patients with Klippel-Trenaunay-Weber syndrome have foot or hand malformations.2 Affected patients often present with pediatric complaints including varicosities, edema, wound healing complications and digital anomalies. As the vast majority of Klippel-Trenaunay-Weber patients are affected in the lower extremity, we present this case in order to help educate the profession on this rare and challenging condition.

Literature review
Klippel and Trenaunay, in their landmark publication described the triad of symptoms listed above, however, they also noted that the clinical manifestations are highly variable. The nevi they identified has since been found to be malformation of underlying capillaries and may or may not take a dermalatrical distribution as originally described.1 76-100% of affected patients presented with visible dilated vessels on the lower leg visible at birth.3 This "Klippel-Trenaunay vein" is joined by other varicosities that become visible as the child grows. Generalized hypoplasia of a hand or foot may be noted by authors as a characteristic finding.2 This includes enlargement of both bone and soft tissue structures causing increased length and girth which may be evident at birth or shortly thereafter.4 Digital anomalies including syndactyly and polydactyly have also been reported.3,4 British dermatologist Frederick Parkes Weber, in 1907, described a condition similar to Klippel-Trenaunay syndrome. In 1918 he described the finding of arteriovenous fistulas in addition to the triad of symptoms previously attributed to Klippel and Trenaunay. Whether this finding necessitates renaming the condition Klippel-Trenaunay-Weber syndrome or if it is a separate entity has been discussed much in the literature.5-7 In general, older publications tend to refer to "Klippel-Trenaunay-Webber syndrome" while more recent literature appears to esteem the two as separate disease entities, "Klippel-Trenaunay syndrome" and "Parkes-Weber syndrome." Previously, authors have noted that the terms "Klippel-Trenaunay-Weber syndrome" and "Klippel-Trenaunay syndrome" appear interchangeably in the literature.5 Regardless of semantics, the disease process originally described by Klippel and Trenaunay may be described in conjunction with arteriovenous fistulas which can cause increased morbidity for the patient.8

Case Report
A 33 year old female with a known diagnosis of right foot polydactyly and Klippel-Trenaunay-Weber syndrome presented to the emergency room with a bullous lesion on her sixth toe. The right foot was enlarged and edematous compared to the contralateral limb. The patient has multiple concomitants including polydactyly, toe malformations, lateral deviation, hypoplasia, and epiphiplasia. She is non-verbal, non-ambulatory and is cared for by her grandmother.

Figure 1: Initial AP radiograph showing post-axial polydactyly

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The sixth toe bullous lesion was lanced and drained in the ER and the patient began an oral antibiotic. The toe regressed and she returned to the emergency room where the superficial non-violent skin was all debrided. This left a large ulceration that encompassed the entire area of the supernumerary digit. There was also significant bleeding noted during the debridement.

Standard wound care methods were employed, however the wound did not improve. Radiographs taken several weeks later showed questionable loss of cortical margin worrisome for osteomyelitis of the distal phalanx. At that point, it was determined that the best, most definitive course would be to proceed with amputation of the sixth toe.

Figure 2: Clinical appearance of the foot prior to wound debridement

The patient continues to struggle with Klippel-Trenaunay-Weber syndrome in addition to multiple other systemic health challenges. Conservative lower extremity treatment is constant, including compression therapy. Unfortunately, the doe experience occasional skin breakdown on the foot but has not, to this point, had a subsequent occurrence of osteomyelitis or severe infection requiring surgery.

Analysis and discussion
Klippel-Trenaunay syndrome is a pathologic entity that is likely not familiar to the majority of practitioners. This is due primarily to the paucity of cases. Prevalence has been reported at slightly less than 1 in 100,000 live births. For comparison sake, the prevalence of talipes equinovarus, a congenital condition much more familiar to the profession, has been reported at 1.4 per 10,000 live births.8 This is more than 100 times the reported prevalence of Klippel-Trenaunay syndrome. As reviewed previously, it is not uncommon for Klippel-Trenaunay patients to manifest digital anomalies, however, cases of polydactyly associated with Klippel-Trenaunay syndrome are rare. All the published cases with the exception of one describe polydactyly with soft tissue involvement only.9,10 The exception does include extra bone formation, however it is a case on the lateral aspect of the left foot.11 While digital anomalies including polydactyly have been described, we were not able to find a case of osseous polydactyly of the foot. Moreover, we did not find recent instances of surgical treatment for the polydactyly associated with Klippel-Trenaunay syndrome. While the nature of the disease necessitates perpetual conservative treatment, the liability of abnormal anatomy may also need to be addressed surgically. This is only recommended after conservative treatments have been exhausted, as seen in the case we have presented here.

Surgical amputation of the sixth toe was successful in removing the ulcer and soft tissue involvement. Copious irrigation was performed and the incision was closed primarily. The surgical wound was slow to heal but did eventually heal completely without complications.

Figure 3: AP radiograph showing loss of cortical definition at the distal phalanx

Figure 4: Intra-operative photograph as the sixth toe is removed

References