Case Report

Klippel Trenaunay syndrome in the context of work-related injury: Case report and review of the literature

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Abstract

Background: Uncommon diseases are usually not suspected at initial presentation, and the diagnosis might be challenging. Here we present a rare disease diagnosed in a work-related injury setting, highlighting the importance of further investigation by means of a more detail physical exam, imaging studies and involvement of other specialties. **Case Presentation:** A 21-year-old Hispanic male, who is a food service worker, presented following a work-related right elbow contusion with severe pain to his right elbow associated with swelling and purplish-red bruising/discoloration on its medial side and forearm. Physical exam demonstrated swelling, tenderness, and conspicuous dilated blood vessels across the right arm and forearm; additionally, multiple red-purplish scattered patches were found on the right arm, anterior and posterior right upper chest. His past medical history was significant for Capillary Hemangioma. He was initially treated conservatively and with work restriction; however, the pain in the forearm persisted. CT angiogram showed multiple interweaving vascular structures on the forearm, and further imaging by MR angiography depicted multiple vascular malformations in the right upper extremity and chest. Vascular surgery was consulted, and the diagnosis of Klippel-Trenaunay syndrome was made. **Conclusions:** Klippel-Trenaunay syndrome is a rare congenital disorder that could present in a wide-range of signs and symptoms. Thorough history taking and clinical examination is warranted in any work-related injuries. Further work up and referral to specialist should always be considered when diagnosis is unclear, or when initial symptoms do not resolve with treatment.

Keywords: Forearm, Klippel-Trenaunay syndrome, rare, work-related

Background

Klippel-Trenaunay (KT) syndrome is a developmental anomaly characterized by the triad of capillary nevus, early onset of varicose veins and tissue hypertrophy of the affected limb. It was first identified by Klippel and Trenaunay in 1900.[1] In 1918, Weber added the association of arteriovenous malformation (AVM) with the syndrome in some cases and hence coined the name Klippel-Trenaunay-Weber (KTW) syndrome when there is arterial involvement. Other names for the syndrome include: angioosteohypertrophy syndrome and hemangiectatic hypertrophy.[2] The syndrome is usually caused by mutations in the *PIK3CA* gene which is involved in the construction of the enzyme PI3K (phosophatidylinositol 3-kinase), cellular proliferation and migration.[3] The syndrome affects about 1 in every 100,000 populations,[3] and lower extremities are affected in 95% of cases. Multiple body parts involvement were also reported in some cases.[2,4] We aim to present this unveiled disease in a work-related injury setting, and the diagnosis course.

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Case Presentation

A 21-year-old Hispanic male was presented to our Occupational Medicine clinic following an injury to his right arm. The patient works as a food service worker and had injured himself while trying to pull a tray out of a tray container when the tray had gotten stuck in the container. The patient lost his control and fell forward, hitting his right elbow with the tray handle. He felt immediate sharp pain in his elbow, associated with swelling and purplish-red bruising/discoloration of the medial side of the elbow and proximal forearm.

The patient reported a history of previous surgical procedure for a benign mass at his right axilla at the age of 4 years. The mass initially was thought to be a lymph node. However, following the procedure, the patient's mother was notified it was a "blood clot" and was removed completely and no need for further investigations.

At age 17, the patient was evaluated by a dermatologist because of a persistent red-purple patch at his right arm and was informed that the patch appears to be a "capillary malformation", for which laser therapy would be an option. However, both parties decided not to perform laser at that point. The patient had uneventful course since then until one month before his recent injury, when he underwent an excisional biopsy for a right axillary lymph node at an outside hospital. He was told it was a "benign" lymph node and that no further treatment or work up was needed.

The patient was seen at our occupational medicine clinic at the same day of the work-related injury. He described his pain level as 8/10 and sharp. There was an obvious bruise and significant tenderness at the medial side of his right elbow and anteromedial proximal forearm. Conspicuous dilated blood vessels were noted across the right arm and forearm. Multiple red-purplish scattered patches were also found on the patient's arm, anterior and posterior upper chest on the right side [Figure 1]. Per the patient, these patches became more prominent after the injury.

The patient was diagnosed with right upper extremity contusion, and was placed on work restrictions of no use of the right arm. An arm sling was dispensed to allow for forearm rest and elevation to decrease the swelling. He was advised to avoid Non-Steroidal Anti-Inflammatory Drugs (NSAIDs) due to the risk of bleeding and was indicated to use Acetaminophen only for pain management. He was also recommended to wear compression sleeves for the right upper extremity. Due to significant worsening of the pain, the patient went to the emergency department and Computed Tomography (CT) angiogram of the right upper extremity was performed. The test showed normal arterial flow but multiple interweaving vascular structures on the forearm. Subsequently, he was discharged from the emergency department to follow up with occupational medicine.

The patient was re-evaluated one week after the injury at the occupational medicine clinic, and his pain level decreased to 6/10 during rest, but occasionally flared up to 8/10 when he moved his right upper extremity. The pain also worsened when he applied ice to the swelling area. Additionally, the swelling, tenderness and discoloration had extended to involve the medial (ulnar) aspect of the distal forearm and hand [Figure 2]. The medial side of the wrist, hand and ulnar 3-digits became tender and cold to palpate. Therefore, he was referred to vascular surgery to address this condition, and the same work restrictions for no use of the right upper extremity were indicated.

By the time of the visit at the vascular surgery clinic, a month later, the clinical condition improved substantially. The pain in the elbow and forearm had significantly decreased and the swelling became less prominent. He continued to wear the compression sleeves of the right upper extremity, and applying heating pads as needed. He had been taking acetaminophen only twice per week. The work restrictions were eased to be: no lifting/pulling/pushing or carrying more than 10 pounds with the right arm.

The diagnosis of Klippel-Trenaunay (KT) Syndrome was suspected. MR angiogram of the right upper extremity and chest wall was requested. Multiple vascular malformations were found in the MRI with the largest one was centered around the anterior musculature of the forearm (8.7 × 3.2 × 5.2 cm) and around the triceps muscle (measured 9.3 × 2.7 × 4.4 cm). Prominent vessels were seen also within the ulna and humerus bones. Prominent vessels were also seen throughout the fifth finger with small regional vascular malformations. Another 2.5 × 1.2 cm lesion was found at the lateral side of the right chest wall [Figure 3].

Because of the patient's persistent pain and the significant size of the vascular malformations, the vascular surgery team decided to perform fluoroscopic-guided sclerotherapy for the vascular

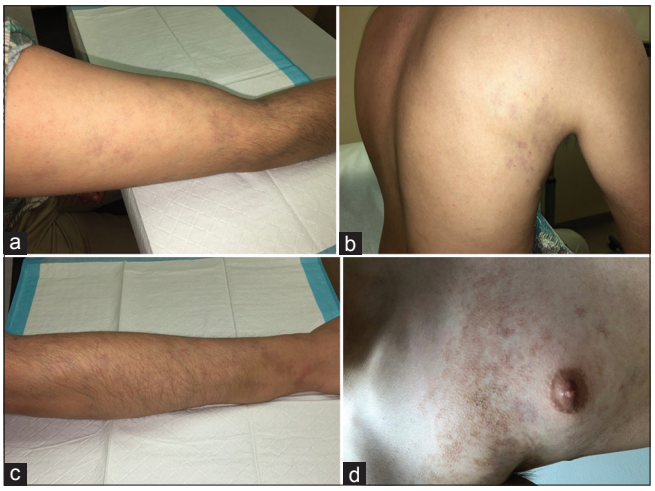

Figure 1: Multiple capillary hemangiomas on the ipsilateral arm (1.a). Multiple capillary hemangiomas on the ipsilateral forearm (1.c). Capillary hemangiomas on the lateral side of the chest wall (1.b). Capillary hemangiomas on the anterior chest wall (1.d)

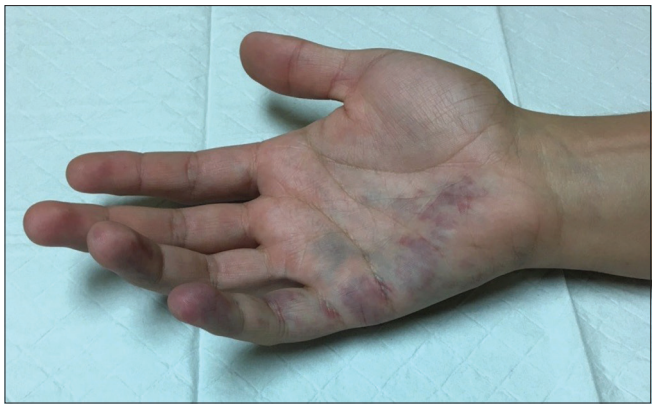

Figure 2: New capillary hemangiomas mainly noted at the ulnar side of the hand

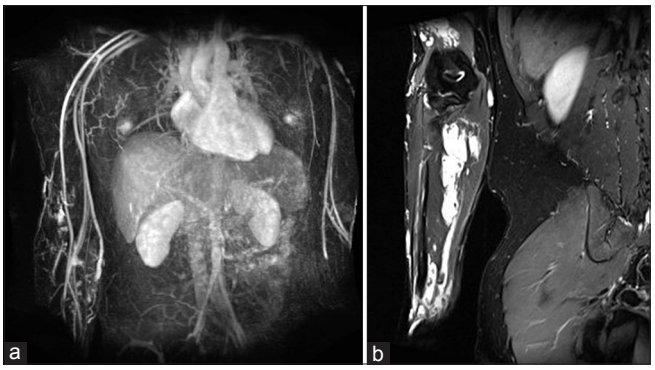

Figure 3: MR-angiogram of the upper extremities shows vascular malformation on the right forearm, arm and right upper chest. No arterial malformation was seen (a). MR-angiogram of the right upper extremity shows multiple arterio-venous malformation of the forearm (b)

malformations of the right upper extremity. Polidocanol 2% was used as sclerosant and the patient tolerated the procedure well.

He was re-evaluated at our occupational medicine clinic, 4 days after the sclerotherapy. He reported slight decrease in the swelling and discoloration in the arm and forearm, however continued to have pain. The patient was placed on further work restrictions of "No lifting, pulling, pushing or carrying more than 5 pounds", following the procedure. 5 days later, the patient presented to the clinic in with worsening pain and firm swelling at the medial side of the arm. There was an excruciating tenderness at that area, and subsequently the Duplex ultrasound was obtained immediately to evaluate for possible deep vein thrombosis (DVT). The ultrasound was negative for any thrombus and only showed post-sclerotherapy changes. The patient reported significant improvement in his pain and reduction of the size of his limb. He was released back to regular duty without any complications.

Discussion

The primary cutaneous manifestation of the KT Syndrome is diffuse superficial capillary malformation that typically presents at the affected limb. When found on the trunk, the malformation rarely crosses the midline and occasionally exhibit sharp demarcation. Vascular malformations usually combine the cutaneous capillary malformation and include abnormal superficial veins, hypoplasia and agenesis of the deep veins, venous duplications and abnormal or dysfunctional valve formation. The limb hypertrophy occurs due to increase in the bulk of the subcutaneous tissues, however, bone hypertrophy can also be observed. Lymphatic vessels abnormalities could also be seen in KT syndrome, which could result in lymphatic obstruction and subsequent worsening of the affected limb swelling. Arterial malformations are rarely seen in KT syndrome.[1,4] The diagnosis of KT syndrome could be achieved accurately using non-invasive procedures. Combination of color Duplex ultrasonography, MRI, lymphoscintigraphy and plain radiographs of bones usually provide sufficient information for the diagnosis and intervention plan. Treatment of KT syndrome involves several modalities. Compression with custom garment has been used for patients with prominent varicosities and edema. Stripping of the varicosities could also be done providing that the deep venous system is intact. Manual lymphatic drainage is a technique used when lymphatic channels are involved and can result in a substantial reduction in the size of the limb. Pulse-dye laser treatment was also used to treat cutaneous capillary malformations and decreasing their associated redness. Fluoroscopic-guided sclerotherapy and embolization are used to treat venous malformations providing that there is no direct connection with the arterial system (to avoid arterial embolization). Eventually, reduction of the limb size could also be achieved surgically by reduction of the bone length (done by orthopedic surgery) which is rarely needed.[1] Complications of KT syndrome are often related to size, shape and location of the abnormal vasculature. These include: thrombosis and pulmonary embolism, pulmonary hypertension due to recurrent pulmonary emboli, heart failure (if presence of a significant AVM), bleeding from involved organs such as gastrointestinal tract, kidneys and genitalia. Infection is a major concern when there is impaired lymphatic drainage. Persistent pain usually occurs due to significant edema and venous insufficiency.[1,4-6]

Our case describes a very rare syndrome that was diagnosed only after further work up initiated by occupational medicine in the context of a work-related injury. Although the patient had prior symptoms and signs and had undergone procedures for vascular abnormalities, these were treated in an isolated fashion and he was never sent for a comprehensive work up to investigate more subclinical lesions and possible causation. Referral to a specialized center had also played an important role in reaching the diagnosis earlier and institution of further necessary treatment. The message from this case is to be aware of any possible rare diseases that present with only common symptoms and signs while hiding many serious subclinical abnormalities which could present in the future with harmful complications. Despite the complexity of the workers' compensation and the insurance system, all the advanced investigations were performed. The referral process and the surgical procedures were done appropriately serving the message of 'patient care comes first'.

Conclusion

Klippel-Trenaunay syndrome is a very rare disorder that could present in a wide-range of symptoms and signs. Thorough history taking and clinical examination is warranted during examination of patients with work-related injury and work-related disorders. Further work up should always be conducted in cases where diagnosis is unclear or symptoms do not resolve in the usual fashion. More importantly, referral to specialist should always be considered when underline causes or associated abnormalities are found, despite the complexity of the workers' compensation insurance system.

Ethics approval and consent to participate

Written informed consent was obtained from the patient for publication of the case report and any accompanying images. Ethical clearance by our institutional review board was not required for publication of the case report of one subject, as protected health information has not been disclosed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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