Rheumatology: Current Research
ISSN: 2161-1149

Gupta, Rheumatology (Sunnyvale) 2018, 8:3
DOI: 10.4172/2161-1149.1000244

Case Report Open Access

Multidisciplinary Approach to Treatment of Severe Klippel-Tranaunay-Weber Syndrome

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Received date: September 22, 2018; **Accepted date:** November 14, 2018; **Published date:** November 29, 2018

Introduction

Klippel-Trenaunay-Weber Syndrome (KTWS) is an extremely rare congenital vascular disorder occurring in less than 1 in 10,000 live births [1]. KTWS consists of a triad of port-wine stain, varicosities or venous malformations, and bony and soft tissue hypertrophy [2]. Approximately 63% of patients have all three features of the syndrome [2,3]. Deep venous abnormalities, brain abnormalities (including AV malformation, aneurysm and atrophy), and hemangiomas of visceral organs including the spleen and bladder are associated with KTWS [4-6]. Pulmonary thromboembolism and hypercoagulability are other rare complications [7]. This can present as a consumptive coagulopathy leading to thrombocytopenia and bleeding, known as "Kasabach-Merritt Syndrome" [8].

Management of KTWS is mainly non-operative, with symptomatic treatment of pain, swelling and bleeding being most common [9,10]. Less than 10% of patients with KTWS undergo vascular surgical treatment (9 out of 144), which include stripping of the veins, and avulsion and excision of varicosities and venous malformations, with 50% recurrence of varicosities [11]. Surgery is typically performed for cosmetic reasons or for chronic venous insufficiency [11]. Symptoms of hematochezia, hematuria and esophageal bleeding are indications for surgical intervention [12]. Lethal complications such as intra-abdominal hemorrhage rupture of lower extremity vessels, and subsequent death can result from delayed treatment of KTWS.

Previous literature reports a maximum leg circumference difference of 6 cm between the affected and unaffected limbs [13,14]. Our patient represents a much more severe presentation of KTWS, with a leg circumference difference of 53 cm, and involvement of the umbilicus through the foot, as well as various visceral organs. This case report will present a possible management approach to severe KTWS using a multi-disciplinary strategy.

Case Presentation

A 44-year old Puerto Rican male with a history of congenital hemangiomas of the right lower extremity (RLE) presented for management of acute high grade fever and severe KTWS. The patient was diagnosed with a primary hemangioma at age two, which stretched from the umbilicus to the right foot, with involvement of the scrotum and perineum. At age four he underwent excision of scrotal hemangioma, followed by radiation therapy at age five, cystoscopy for bladder hemangioma at age 13, and resection of partial bleeding hemangioma of the RLE at age 21. The patient did not seek further treatment until age 38 when he underwent splenectomy for large hemangioma of the spleen, discovered incidentally after motor vehicle accident. The patient presented to our institution at age 44 with symptoms of infection and anemia, with his hemangioma draining blood.

Physical examination of the patient revealed a port-wine stain measuring 4 cm × 4 cm on the outer right thigh, shortening of the RLE, and a deformed right foot. The right thigh measured 93 cm at greatest circumference compared to 40 cm for the left thigh. The RLE, buttocks and testicle were swollen and erythematous. The patient ambulated with crutches, and had lost all function of the right lower limb. Additional findings included a dense tissue mass on the right breast, and multiple small hemangiomas on the chest wall measuring <0.5 cm[15].

The patients PMH was significant for cardiac enlargement diagnosed as a child and chronic hepatitis C. Medications included only Tramadol HCl for hemangioma-related pain relief. Family history was significant for multiple cherry hemangiomas in his mother, a vague history of the mother experiencing pulmonary "blood vessel rupture," and a strong positive matrilineal history of varicose veins. The patient had three unaffected siblings and reported no early deaths or any relatives. The patient had a 40-pack-year history of cigarette smoking.

Hospital Course

The patient initially presented with one week of fevers (Tmax 103°F), associated with increasing erythema and tenderness of the upper RLE. Symptoms resolved with a course of vancomycin and keflex for 10 days, to treat skin structure infections. The patient also requested a more definitive management his RLE hemangioma, as the increasing size was severely impeding his quality of life. Initially multiple services were consulted regarding surgical management of the hemangioma. Hematology was consulted due to the documented complications of coagulopathies in KTWS, particularly Kasabach-Merritt [8]. CT of the chest showed large L mediastinal cystic structure, and numerous diffuse bilateral pulmonary nodules suggestive of pulmonary metastasis [16]. Oncology recommended tissue biopsy due to concern of possible angiosarcoma of the RLE (potentially related to remote radiation therapy history) with possible metastasis to the lung. A biopsy of a posterior pulmonary nodule showed no evidence of malignancy. MRI/MRA of RLE showed a large hemangioma, with multiple discrete solid components. These components were also hypermetabolic on PET/CT.

Hematology advised that the patient would likely require anticoagulation immediately before undergoing any surgery due to a risk of Kasabach-Merritt. Orthopedic surgery concluded that the morbidity of limb-sparing surgery would be unacceptably high. Based on the patient's lack of functionality of the RLE, orthopedic surgery determined that the most prudent approach was likely amputation of

the extremity with hemipelvectomy given the extent of the vascular malformation invading the pelvis. At an interdisciplinary meeting, the medical teams judged that immediate surgical management could involve significant blood loss and morbidity, and that embolization prior to surgery would be the safest approach. Interventional radiology and vascular surgery considered doing a single embolization of the major arterial feed immediately prior to surgery,but due to the size and complexity of the vascular malformation, they concluded that a staged embolization to progressively decrease the size and vascularity of the limb would be optimal to maximize both the ongoing safety of the patient and the feasibility of the planned surgery. Due to the unusual nature of the case, embolization was deferred to a local interventional cardiologist specializing in high risk embolizations[17].

Conclusion

This case illustrates the evolving complexity of an extreme presentation of a rare condition. Due to the differing decisions of individuals about when to seek medical care and to what extent, it is sometimes necessary for medical teams to confront novel management issue which lack an accepted protocol. In this case, we believe that the patient not only represents the most extreme case of KTWS documented, using limb size discrepancy as a metric, but also that he illustrates how different aspects of a condition require triage by the primary medical team. Multiple medical and surgical specialty teams were able to devise a tailored approach by communicating directly with each other and the patient so that his personal priorities could be used as a unifying focus. Although staged embolization followed by surgery has been documented as a successful approach to isolated arterio-venous malformations, to our knowledge this represents the first time that it has been considered for a syndromic hemangioma of this complexity. This case also reminds us that it remains essential to treat KTWS hemangiomas as early as possible, due to an almost unlimited increase in potential morbidity of the hemangiomas over time, especially when other risk factors for malignancy are present, such as a history of radiation exposure.

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