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Case report

A case of recurrent massive pulmonary embolism in Klippel–Trenaunay–Weber syndrome treated with thrombolytics

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ABSTRACT

Klippel – Trenaunay – Weber syndrome (KTWS) is a congenital condition characterized by a triad of capillary malformations of the skin, soft tissue and bone hypertrophy resulting in limb enlargement, and abnormalities of arteriovenous and lymphatic systems of the affected limb. In this case, we present a patient with KTWS receiving chronic anticoagulation that had a massive pulmonary embolism and was successfully treated with thrombolytic therapy. The purpose of this case is to educate readers about this uncommon condition and to increase awareness, recognition and timely treatment of its most common complications, namely thrombosis and pulmonary embolism.

1. Introduction

Klippel–Trenaunay–Weber syndrome (KTWS) is a rare congenital condition characterized by extensive venous malformations and soft tissue and bone hypertrophy. KTWS predisposes to hypercoagulable states, including venous thromboembolism (VTE) and pulmonary embolism (PE). The incidence of PE ranges from 14 to 22%; however, recurrent massive PE is unusual. We report the case of an adult female with KTWS who developed recurrent massive PE and was successfully treated with thrombolytic therapy.

2. Case presentation

A 36 year old Guyanese female with a history of KTWS and previous massive PE one year prior, noncompliant with anticoagulation therapy, presented with sudden onset pleuritic chest pain, dyspnea and diaphoresis. She denied nausea, vomiting or cough. At presentation blood pressure was 80/64 mmHg, pulse was 150 bpm, respiratory rate was 22 and SpO2 on room air was 87%. Physical examination revealed significant swelling of the left leg with prominent venous irregularities.

Admission labs revealed elevated troponin-I (0.430 ng/ml), D-dimer (9056 ng/ml) and BNP (1280 Pg/ml) levels with subtherapeutic INR (1.2) (See Table 1 for further details on admission labs). Arterial blood gas showed high A-a gradient hypoxia.

EKG showed sinus tachycardia at 144 bpm with non-specific ST–T changes.

Chest radiogram failed to show any gross infiltrate, atelectasis or effusion (Fig. 1). CT pulmonary angiogram revealed filling defects in both the pulmonary arteries (Fig. 2A and B) with no pulmonary infarction. Filling defects extended to the lobar branches distally.

2D echocardiogram showed increased right ventricular size and bowing of the septum into the left ventricle (Fig. 3) with elevated PA pressure (64 mmHg). Vascular venous doppler of the legs confirmed the presence of deep vein thrombosis in the left leg (Fig. 4).

Thrombolytic therapy was administered with improvement in hemodynamic status over the next 24 h. Repeat 2D echocardiogram showed resolution of right ventricular overload. The patient was advised to continue lifelong anticoagulation therapy.

3. Discussion

The common pulmonary manifestations of KTWS include thromboembolic phenomena, pulmonary venous varicosities, pulmonary lymphatic obstruction and cavernous hemangiomas of the pleura [1,4]. The etiology and pathogenesis of capillary

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Table 1
Pertinent admission laboratory data.

WBC count	7100/mm^3
Hemoglobin	14.5 gm/dL
Hematocrit	44.1%
Prothrombin time	13.3 s
INR	1.2
PTT	22.5 s
Blood urea nitrogen	23 mg/dl (H)
Creatinine	0.5 mg/dl
Sodium	137 mEq/L
Bicarbonate	24 mEq/L
Chloride	104 mEq/L
Troponin	0.430 ng/ml (H)
BNP	1280 Pg/ml (H)
D-Dimer	9056 ng/ml (H)
Arterial blood gas on room air	7.44/29(L)/64(L)

*Note: (H) indicates high, (L) indicates low.

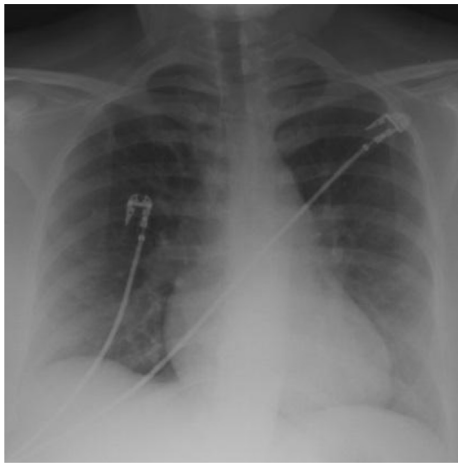

Fig. 1. X-ray showing no infiltrate, atelectasis or effusion.

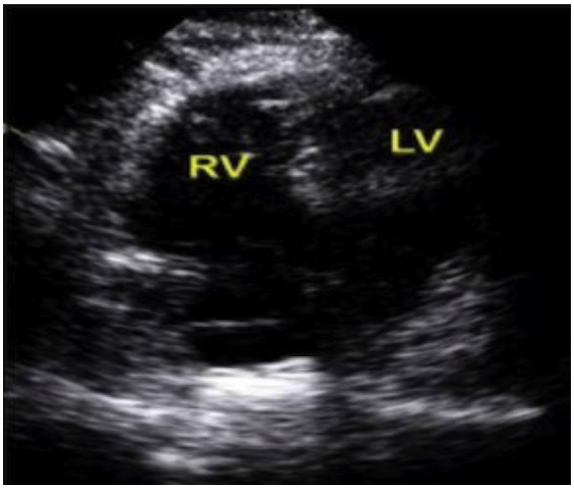

Fig. 3. Apical four chamber view of 2D Echo revealing bowing of the inter-ventricular septum and right-sided enlargement (RV/LV > 1) suggestive of massive pulmonary embolism.

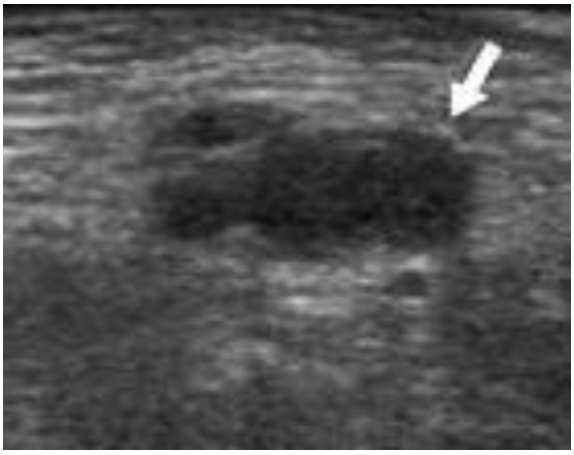

Fig. 4. Venous Doppler showing lack of compressibility of the left femoral vein (white arrow) suggestive of the presence of deep vein thrombosis.

malformation are yet to be fully elucidated; however, several mechanisms have been proposed, including vascular ectasia, lack of neuronal control of blood flow, as well as involvement of a vascular endothelial growth factor and its receptor [2].

Vascular malformations of capillaries, arteries and veins associated with this syndrome are present at birth and do not regress, but actually become more prominent with time [3]. Patients with this condition have gross hypertrophy and overgrowth of soft tissues and bones of the affected limb. The affected skin is thickened, nodular, consistent with a cobblestone appearance, and prone to develop ulcers. The lower limb is affected significantly

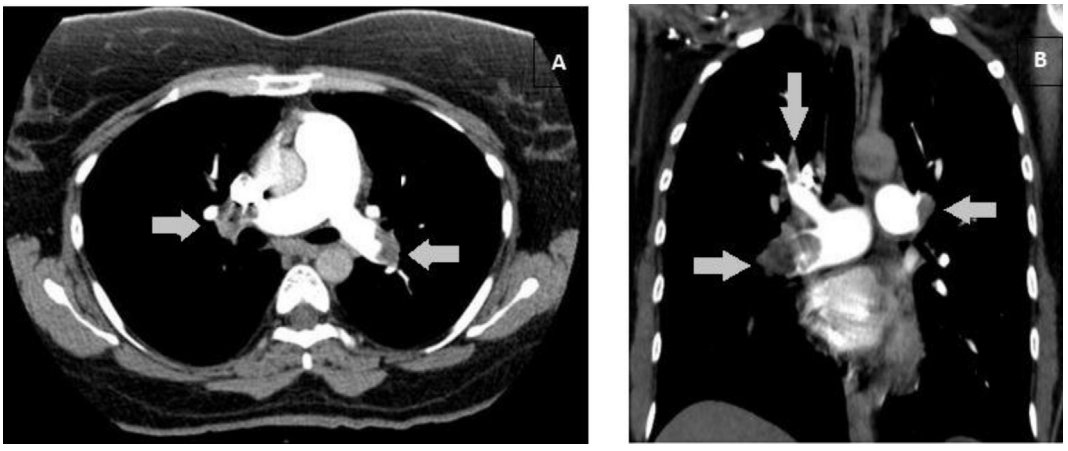

Fig. 2. (A and B): CTPA showing pulmonary emboli in bilateral pulmonary arteries (Arrows).

more often (95%) than the upper limb (5%) or both limbs (15%) and the majority of the cases have a unilateral involvement [5].

For an improved cosmetic appearance, treatment of capillary malformation includes pulsed dye laser therapy [2]. The marked hypertrophy and length discrepancy of the affected limbs necessitate orthopedic intervention when feasible. Patients with KTWS are at risk of developing high output cardiac failure. Increased propensity for thrombosis and emboli make lifelong anticoagulation therapy a necessity [2,6].

Missed diagnosis of these KTWS associated thrombotic and cardiac complications can potentially be life threatening. Due to the rarity of this disease, there are no recommended guidelines for the management of thromboembolic phenomena. However, aggressive therapeutic anticoagulation and an early placement of IVC filter have been suggested [1,2].

4. Conclusion

Klippel–Trenaunay–Weber syndrome is a rare congenital condition and associated complications can at times be difficult to manage. The management and prophylaxis for the recurrence of pulmonary embolism and other thromboembolic events are the areas that need special attention.

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