Pleural effusion in Klippel–Trenaunay syndrome: an uncommon manifestation

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Abstract

Klippel–Trenaunay syndrome is characterized by a combination of vascular abnormalities and limb hypertrophy. Pleural effusion as a manifestation of this syndrome is almost never mentioned in the literature. We present a case of persistent bilateral pleural effusion in a patient with Klippel–Trenaunay syndrome and share our experiences treating this scenario.

Keywords: Klippel–Trenaunay syndrome • Pleural effusion • Pleurodesis

INTRODUCTION

The combination of vascular abnormalities and limb hypertrophy was first reported in 1900 by 2 French doctors, Klippel and Trenaunay [1]. Later known as the Klippel–Trenaunay syndrome (KTS), this condition is a combination of overgrowth and low-flow vascular malformation and includes varicosities, tissue hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2]. The genetic base of this syndrome is related to the presence of a somatic mutation in the phosphatidylinositol 3-kinase (PIK3CA) gene, leading to cellular hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2]. The genetic base of this syndrome is related to the presence of a somatic mutation in the PIK3CA gene, leading to cellular hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2]. The genetic base of this syndrome is related to the presence of a somatic mutation in the PIK3CA gene, leading to cellular hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2]. The genetic base of this syndrome is related to the presence of a somatic mutation in the PIK3CA gene, leading to cellular hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2]. The genetic base of this syndrome is related to the presence of a somatic mutation in the PIK3CA gene, leading to cellular hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2]. The genetic base of this syndrome is related to the presence of a somatic mutation in the PIK3CA gene, leading to cellular hypertrophy and cutaneous port-wine stains on the affected limb(s). Mostly the lower limbs, but also upper limbs or truncal involvement was described. Today, lymphatic anomalies are also considered a part of the syndrome [2].

CASE PRESENTATION

A 25-year-old man diagnosed with KTS was referred to our department for evaluation due to accumulation of bilateral pleural effusions causing severe dyspnoea. The patient had been previously treated repeatedly with pleurocentesis and bilateral chest tube drainage but still had persistent reaccumulation of the effusion. Upon admission, large bilateral pleural effusions were demonstrated on thoracic imaging. Bilateral thoracic chest drains were placed; the daily production of effusion was >1000 cc.

To further assess the disease, we first performed left video-assisted thoracoscopic surgery (VATS). Surgical findings were mainly a collapsed lung with a relatively thin peel on the visceral pleura and some fibrin deposits. Pleural pathology showed chronic follicular pleuritis. A silicon 28-Fr chest drain and a tunneled pleural catheter drainage system (PleurX, Becton, Dickinson, Franklin Lakes, NJ, USA) were put in place and chemical pleurodesis using doxycycline was applied. The silicon drain was removed after a few days, and pleural drainage was continued using the tunneled pleural catheter system for 5 months until no more fluid accumulated and the lung re-expanded.

Ten days after the first procedure, we performed a VATS evaluation of the right pleural cavity. Chronic adhesions resembling an empyema-like complex pleural space with partly trapped lung were noted. After decortication and lung re-expansion, chemical pleurodesis was applied. A silicon 28-Fr chest drain was placed in the pleural cavity at the end of the operation. The drain was removed on postoperative day 8. On follow-up, reaccumulation of effusion and right lung atelectasis were noted. A tunneled pleural catheter drainage system was inserted 1 month after VATS decortication and pleurodesis. In the following months, the right pleural fluid subsided and the lung gradually re-expanded. The drainage system was removed 4 months after its insertion.

At the 3-year follow-up, the patient was free of respiratory symptoms with no evidence of the reaccumulation of effusion.

COMMENT

Patients with KTS suffer from a wide variety of symptoms related to their deformed and overgrown extremity such as a tendency to bleed, localized intravascular coagulopathies and veno- thromboembolic events.
The presence of persistent embryonic veins (lateral marginal vein and sciatic vein) represents vascular malformations mostly seen in patients with KTS.

Lymphatic malformations are also common with dermal microcystic and pelvic macrocystic expression.

Lung involvement in patients with KTS is mostly manifested as pulmonary arterial hypertension due to recurrent pulmonary embolism, which is attributed to the tendency to have localized intravascular coagulopathies [2].

Pleural effusion as a symptom of KTS is almost never mentioned in the literature. A literature search revealed only 1 reference to this symptom by Joshi et al. [4]. The authors described a case of a young adult with known KTS who developed pleural and pericardial effusions. A lung biopsy revealed lymphangiectatic sclerosis with smooth muscle hyperplasia that they believed represented lung involvement in patients with KTS. The presence of pleural effusion was attributed to the abnormal lymphatic drainage with obstruction and hyperplasia of pre-existing lymphatic channels.

They treated the patient with repeated thoraco- and cardiocentesis, which failed to clear the effusion. There are no data related to pleural fluid reaccumulation after they performed a lung biopsy and pericardiectomy [4].

Our case focuses on the clinical approach to the unique scenario of pleural effusion in patients with KTS. Because repeated pleurocenteses were futile in clearing the pleural cavity, we chose an integrated therapy. We first performed thoracoscopy with chemical pleurodesis. Considering our inability to predict the success of pleurodesis in patients with KTS, we also placed a tunneled pleural catheter drainage system. This strategy was beneficial in view of chemical pleurodesis failure in the left pleural space. The use of the tunneled pleural catheter continued until the accumulation of fluid subsided and the lung re-expanded (>5 months).

Right-sided thoracoscopy findings of the complex pleural space dictated the execution of lung decortication resulting in lung re-expansion followed by chemical pleurodesis. We believed that this approach would be sufficient, but as occurred with the other side, the pleural effusion reaccumulated. Only after inserting a tunneled pleural catheter drainage system were we able to resolve this complex pleural situation with full lung re-expansion after 4 months.

This is the first description in the literature of this treatment approach to pleural effusion in patients with KTS. Our experience shows that chemical pleurodesis is futile and that the use of a tunneled pleural catheter drainage system is a suitable solution for this scenario.

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