

## CASE REPORT

# TUBEROUS SCLEROSIS – RARE PRESENTATION AS PNEUMOTHORAX

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Pulmonary disease is a rare manifestation of Tuberous Sclerosis. We report a case where the patient presented with seizures, shortness of breath, popular lesions on the face and warty growths over the skin. She was intubated and the lung expanded on the 7<sup>th</sup> day after a longstanding pneumothorax of 3 months.

**Key Words:** Pneumothorax, Tuberous Sclerosis, Seizures.

## INTRODUCTION

Tuberous sclerosis is an autosomal dominant disorder characterized by mental retardation, epilepsy, tumours of skin, kidney, heart, brain and eyes.<sup>1</sup>

Pulmonary involvement is very uncommon and affects women of child bearing age.<sup>2</sup> There are diffuse interstitial infiltrates and cystic changes resulting in recurrent pneumothorax.<sup>3</sup> Most patients of tuberous sclerosis with chest involvement have a slowly declining clinical course resulting in cor pulmonale and pneumothorax.<sup>3</sup>

## CASE DESCRIPTION

A 33 years old lady was admitted in Pulmonology unit of the Post Graduate Medical Institute, Lady Reading Hospital Peshawar on September 6, 1999 with the complaints of dry cough, fever off and on and left-sided chest pain for the last five months. The pain started suddenly and was not related to exertion. There was no history of haemoptysis or weight loss. Her appetite was good. She used to get breathless on exertion. She gave a history of repeated attacks of epileptic fits since childhood. She consulted many physicians and the diagnosis of Pulmonary Tuberculosis with epilepsy was made for which she received treatment. She did not improve with the treatment.

On physical examination, her pulse was 96 beats per minute and her blood pressure was 120/80 mm Hg. She was not anaemic and her temperature was normal. Her JVP was normal and lymph nodes were not enlarged. She was not cyanosed.

Her chest examination revealed hyper resonant percussion note on the left side and breath sounds were decreased on the same side. There were no added sounds. Examination of other systems did not disclose any abnormality.

**Figure-1: There were reddened nodules on the cheeks, nasolabial folds, sides of the nose and chin.**

**Figure-2: Examination of the digits revealed subungual fibromas prominent on the feet.**

**Figure-3: Fundal examination showed hypopigmented retinal patches**

**Figure-4: Chest X-rays revealed Pneumothorax of the left lung**

**Figure-5:CT scan of the brain showed paraventricular calcification**

**Figure-6: CT abdomen showed multiple hamartomas in the kidneys.**

#### **Figure-7: Lung got expanded in 6 days without any complications**

Her laboratory investigations showed an Hb level of 12.4 g/dl, a fasting blood sugar level of 100 mg/dl and serum calcium of 9 mg/dl. Renal function tests were normal, serum electrolytes were normal, ECG and Echocardiography were normal.

The diagnosis of Tuberous Sclerosis with left-sided pneumothorax was made. She was intubated and the lung got expanded in 6 days without any complications (Figure 7).

#### **DISCUSSION**

Tuberous Sclerosis is an autosomal dominant disorder of variable penetrance<sup>1</sup> characterized by epilepsy, mental retardation, adenoma sebaceum, subungual fibromas, depigmented Ash-leaf lesions of skin and multiple hamartomas in Brain, Kidneys and Heart and Eyes. Males are affected more than females.<sup>2</sup>

Pulmonary involvement in tuberous sclerosis is very rare<sup>4</sup> and seems to be associated with a more benign course.<sup>3</sup> The patients with lung involvement differ from others in that they are predominantly females<sup>2</sup> and are of normal intelligence, as noted also in this case report. Those affected present with recurrent pneumothorax due to recurrent cyst formation in the lung, because of fibrous tissue degeneration. Associated restrictive and obstructive changes occur in pulmonary function. There is a progressive decline in lung function resulting in respiratory failure and cor pulmonale, requiring heart-lung transplant.<sup>6</sup>

This patient was treated with chest intubation and oxygen therapy. She made an uneventful recovery within a week and no recurrence has been reported so far, thus running a benign course, as reported in the literature.

## **CONCLUSION**

Tuberous Sclerosis may very rarely present as pneumothorax. It is commonly seen in women of childbearing age and runs a benign course as compared to tuberous sclerosis without pulmonary involvement.

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