A case presentation with bilateral Pneumothoraces secondary to tuberous sclerosis

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Declaration of conflicting interests

The Authors declares that there is no conflict of interest.

Abstract

Bilateral pneumothoraces is a rare presentation. Here we discuss a case with Bilateral spontaneous pneumothoraces secondary to Tuberous sclerosis. Tuberous sclerosis is a rare autosomal dominant disease characterized by numerous benign tumors in different parts of the body caused by mutations in either of two different genes ,TSC1 and TSC2.

We had a 16 years old girl presented with bilateral spontaneous pneumothoraces. She had a history of left nephrectomy for angiomyolipoma (AML) of left kidney at the age 13 and an episode of fits at the age of 1 month. On examination she had multiple papules on face and neck (adenoma sebaceum).High resolution Computed Tomography (HRCT) chest showed bilateral multiple small cysts and pneumothoraces both sides. She was managed with prompt chest intubation bilaterally for lungs re-expansion, oxygen therapy, intravenous antibiotics and chest physiotherapy with resolution of the pneumothoraces. nical presentation, in particular IgG4-Related disease (IgG4-D) is not clear.

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Introduction

Tuberous sclerosis is a rare ,multi-systemic autosomal dominant disorder that causes benign tumors in various organs of the body mostly involving brain, kidney ,lungs ,heart and skin . It is caused by mutations in either of the genes TSC1 or TSC2, which encode for the proteins hemartin and tuberin respectively. These proteins are tumor growth suppressors that regulate cell proliferation and differentiation. The live birth prevalence is estimated to be 10 and 16 cases per 100,000.[1-3] A study in 1998 estimated total population prevalence between 7 -12 cases per 100 live births. There is no definitive treatment, but medicine, education and occupational therapies decrease the symptomatology of the patients.[1-4]

Case report

A 16 years old girl ,resident of Mardan, presented to Pulmonology department, Lady Reading Hospital Peshawar with sudden onset of shortness of breath, severe chest pain and dry cough for one day. She has significant past history of left sided nephrectomy for angiomyolipoma (AML) . She had an episode of fits at the age of one month for which she was started on antiepileptic but discontinued later as she remained fits free for the rest of her life.

On physical examination she was short of breath and there was significant decreased air entry bilaterally and was saturating 86 percent on room air. There were multiple small red-brown papules over bridge of nose, over the face and lower part of the neck as well as on upper part of the chest on right side which was consistent with adenoma sabaceum (figure 1). Nervous system and cardiovascular examinations were normal.

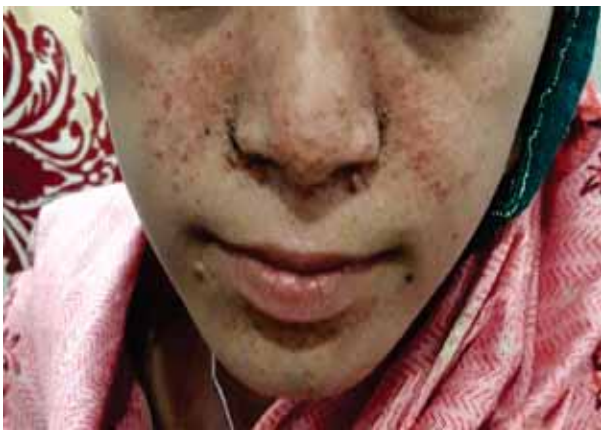

Figure 1: multiple small papules on face since childhood (Adenoma sebaceum).

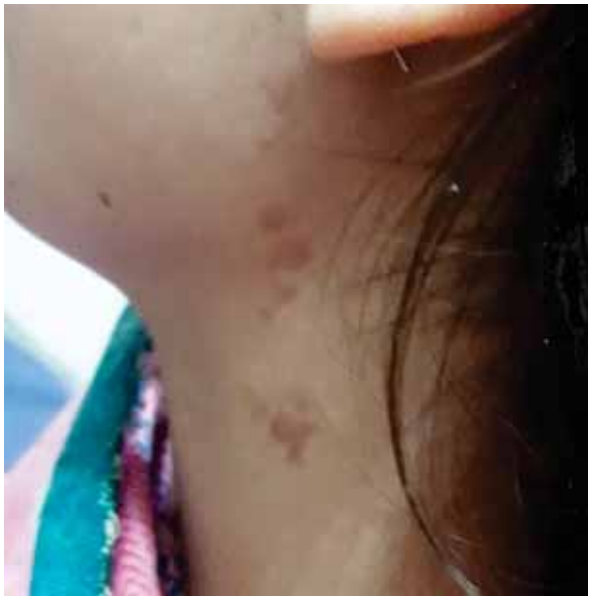

Figure 2: Rash on left lower jaw and upper part of neck.

Investigations

Her chest x ray showed bilateral pneumothoraces (figure 3) with normal leukocytes count, normal renal function tests and normal liver function tests. HRCT showed bilateral pneumothoraces with multiple cysts/bulla more on the right lung parenchyma largest one measuring 18mm in right lower lobe (figure 4). Ultrasound abdomen showed absence of left kidney and normal right kidney.

Figure 3: Chest radiograph showing bilateral spontaneous pneumothoraces with bilateral chest tube in situ.

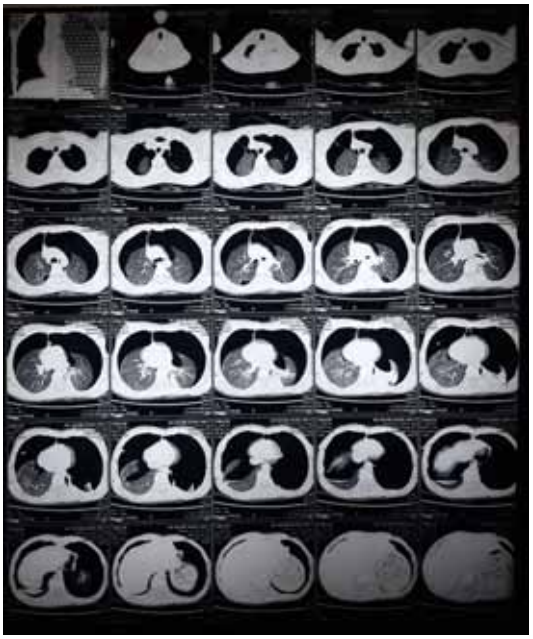

Figure 4: High-resolution CT scan of the chest showing marked pneumothoraces.

Diagnosis

Keeping her clinical presentation, significant past history, extensive physical signs and investigations, a diagnosis of Tuberous Sclerosis were made.

Management and Outcome

The acute treatment of this patient included urgent tube thoracostomy bilaterally and high flow oxygen for lung re-expansion. Symptomatically she improved with time following lung re-expansion and chest physiotherapy. She was discharged after chest tubes removal with advice to avoid strenuous exertion and flights. She was reviewed on regular follow ups with mild shortness of breath on exertion which was relieved with inhaled bronchodilators.

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